Chapter 3: Non-Invasive Prenatal Testing

A. Non-Invasive Prenatal Testing in Germany

I. NIPT in the Private Sector

The first non-invasive prenatal test (NIPT) available in Germany was marketed in 2012 under the trade name PraenaTest by the company LifeCodexx. Under § 3(1)(b) of the former Medical Devices Act (*Medizinproduktegesetz*, MPG)[1420] the test qualified as a medical device for the detection of disability and therefore only required a CE mark to be placed on the market in Germany.[1421] Its placing on the market immediately sparked considerable controversy and public debate. After a series of articles denouncing the market entry of the test in national newspapers[1422] a legal expert opinion commissioned by the Federal Government Commissioner for Matters relating to Persons with Disabilities was released.[1423]

The opinion, drafted by Klaus Ferdinand Gärditz, argued that the test could not lawfully be placed on the market.[1424] According to § 4(1) of the old MPG it was prohibited to place a medical device on the market when there were reasonable grounds for suspecting that they directly or indirectly endangered the safety and health of patients or third parties. In the legal expert's view the medical device legislation would lead to a ban on the marketing of NIPT and an obligation on the competent authorities to prevent it from being placed on the market[1425] because the foetus was

1420 The Medical Devices Act was replaced in May 2021 by the Medical Devices Implementation Act (*Medizinprodukterecht-Durchführungsgesetz*, MPDG).

1421 According to the then current § 6(1) MPG, as pointed out by Huster, 'Der Gemeinsame Bundesausschuss als Ethikbehörde?' (2017) 35(4) MedR p. 282, 283.

1422 As reported by Braun and Könninger, 'Realizing Responsibility.: Institutional Routines, Critical Intervention, and the "Big" Questions in the Controversy over Non-invasive Prenatal Testing in Germany' (2017) 37(3) New Genetics and Society p. 248, 256.

1423 Gärditz, 'Gutachtliche Stellungnahme zur Zulässigkeit des Diagnostikprodukts "PraenaTest"' (2012), pp. 10-11. <https://cdl-online.net/uploads/pdf/praenatest.pdf> accessed 28.9.2021

1424 ibid, p. 11.

1425 ibid.

argued to be a third party under the MPG. Its health and safety would then be endangered because there was a 90% chance of it being aborted as a result of the information revealed by the test.[1426]

The competent authorities did not act in accordance with this opinion.[1427] Moreover, the expert's assessment was contradicted by a subsequent opinion of another legal expert appointed by the manufacturer[1428] and by contributions from other legal scholars.[1429] The second legal expert's opinion found that the foetus could not be regarded as a third party within the meaning and the spirit of the Medical Devices Act.[1430] Furthermore, the opinion argued that the decisive factor in this respect is the fact that the mere use of the test poses no danger to the health and safety of the foetus.[1431] The test discloses information that, in itself, could also be beneficial in protecting the health of the unborn child, for example by choosing appropriate delivery methods for a genetically affected foetus.[1432] By contrast, the possibility that the foetus might suffer harm to its health as a result of the information provided by the test would depend entirely on the mother's decision to have an abortion.[1433]

The legal and ethical controversies that followed the introduction of NIPT tests onto the market also prompted the Federal Government to seek the opinion of the German Ethics Council.[1434] The Council assumed that,

1426 ibid, p. 5.

1427 As notices Huster, 'Der Gemeinsame Bundesausschuss als Ethikbehörde?' (2017) 35(4) MedR p. 282, 283.

1428 Hufen, 'Zur verfassungsrechtlichen Beurteilung frühzeitiger pränataler Diagnostik: Dargestellt am Beispiel des Diagnoseprodukts PraenaTest®' (4.1.2013) <https://lifecodexx.com/wp-content/uploads/2015/03/Jan-2013_PraenaTest_Zur_verfassungsrechtlichen_Beurteilung_fruehzeitiger_praenataler_Diagnostik_Friedhelm_Hufen.pdf> accessed 21.9.2021.

1429 *Inter alia,* Huster, 'Der Gemeinsame Bundesausschuss als Ethikbehörde?' (2017) 35(4) MedR p. 282, 283; Huber in Steger, Orzechowski and Schochow, *Pränatalmedizin: Ethische, juristische und gesellschaftliche Aspekte* (2018) pp. 148-ff.

1430 Hufen, 'Zur verfassungsrechtlichen Beurteilung frühzeitiger pränataler Diagnostik', 4.1.2013, p. 9.

1431 ibid, p. 10, in contrast to the previously used invasive procedures, which, as indicated above in Chapter 1, sec. A.I.3.b pose a small risk of miscarriage.

1432 Huster, 'Der Gemeinsame Bundesausschuss als Ethikbehörde?' (2017) 35(4) MedR p. 282, 283 mentions, for instance, the possibility of choosing a caesarean section rather than natural birth.

1433 Huster, 'Der Gemeinsame Bundesausschuss als Ethikbehörde?' (2017) 35(4) MedR p. 282, 283; Huber in Steger, Orzechowski and Schochow, *Pränatalmedizin* (2018) p. 149.

1434 Deutscher Ethikrat, 'The Future of Genetic Diagnosis' (2013) p. 7.

since the authority responsible for reviewing the test did not object to its marketability under the Medical Devices Act, the test was legally placed on the market.[1435] On the basis of this premise the majority of the Council's members focused on envisaging a legal framework for an ethically acceptable use of NIPT. This would for instance include appropriate and comprehensive information and counselling[1436] and limiting the possibilities for performing NIPT to cases of pregnancy with an increased risk of genetic conditions in the foetus.[1437]

II. NIPT in the Statutory Health Insurance

1. Access to Prenatal Testing

a Prenatal Diagnoses in the Statutory Health Insurance

The reimbursement of prenatal diagnoses by the statutory health insurance is regulated in § 24d SGB V according to which the insured subject is entitled to medical care and midwifery assistance during pregnancy, including prenatal care. The medical services to be offered during pregnancy are specified in the guidelines on medical care during pregnancy and after delivery (*Richtlinien über die ärztliche Betreuung während der Schwangerschaft und nach der Entbindung*, Mu-RL), or Maternity Guidelines, issued and updated by the Federal Joint Committee.[1438]

According to the maternity guidelines a primary objective of prenatal care is the early detection of high-risk pregnancies and births.[1439] For this purpose the pregnant woman is entitled to a number of examinations, including early detection and investigation of risk pregnancies. Not included in the statutory health insurance offer is the so-called first-trimester screening, which is a combined blood test and ultrasound examination procedure

1435 ibid, p. 80.
1436 ibid, pp. 157-158.
1437 ibid, p. 165.
1438 Pursuant to § 92(1) sentence 2 no. 4 of the SGB V, see Welti in Becker and Kingreen, *SGB V: Gesetzliche Krankenversicherung Kommentar* (7th edn 2020) para. 1.
1439 Gemeinsamer Bundesausschuss (G-BA), Mutterschafts-Richtlinien, Richtlinien über die ärztliche Betreuung während der Schwangerschaft und nach der Entbindung 10.12.1985, p. 2.

that can estimate the probability of a chromosomal trisomy being present. The first-trimester screening can be conducted between the 11th and 14th week, but the cost must be borne out-of-pocket.[1440]

If this initial screening gives indications that there may be a trisomy, the pregnancy is classified as at risk. In such instances the statutory health insurance covers the costs of an invasive diagnosis such as amniocentesis or chorionic villus sampling.[1441] A pregnancy is automatically considered to be at risk – and non-invasive diagnoses are therefore reimbursed even without a previous first trimester screening – when the woman is a first-time mother and over 35.[1442]

The prenatal invasive diagnoses thus offered by the statutory health insurance are seen as controversial by some legal scholars[1443] who argue that the aim of medical care during pregnancy is to avoid dangers to the life and health of the child and not the early detection of disabilities that might lead to an abortion. This is considered to be an explanation for the lack of reimbursement for the first trimester screening.[1444] However, it does not explain the statutory health insurance's coverage of possible abortion procedures.[1445]

Against the background of this existing discussion, the emergence of NIPT has sparked debates among legal and ethics scholars on its possible reimbursement by health insurance funds.[1446]

1440 Huber in Steger, Orzechowski and Schochow, *Pränatalmedizin* (2018) p. 145; Kießling in Rolfs and others, *BeckOK Sozialrecht* (61st edn 2021) para. 8.

1441 Gemeinsamer Bundesausschuss (G-BA), Mutterschafts-Richtlinien 10.12.1985, p. 10; Kießling in Rolfs and others, *BeckOK Sozialrecht* (2021) para. 8.

1442 Gemeinsamer Bundesausschuss (G-BA), Mutterschafts-Richtlinien 10.12.1985, pp. 9-ff; Kießling in Rolfs and others, *BeckOK Sozialrecht* (2021) para. 10.

1443 Welti in Becker and Kingreen, *SGB V* (2020) para. 4.

1444 Huber in Steger, Orzechowski and Schochow, *Pränatalmedizin* (2018) p. 145; Welti in Becker and Kingreen, *SGB V* (2020) para. 4; Kießling in Rolfs and others, *BeckOK Sozialrecht* (2021) para. 9.

1445 Found in compliance with the Basic Law by the BVerfG in its second abortion decision (BVerfG, 28.5.1993 - 2 BvF 2/90, 2 BvF 4/90, 2 BvF 5/92, BVerfGE 88, 203), see Kießling in Rolfs and others, *BeckOK Sozialrecht* (2021) para. 10.

1446 See, *inter alia*, Huster, 'Der Gemeinsame Bundesausschuss als Ethikbehörde?' (2017) 35(4) MedR p. 282; Buyx, 'Kostenübernahme für pränatale Bluttests. Pro und Contra' (2018) 115(44) Deutsches Ärzteblatt A1988; Rüffer, 'Kostenübernahme für pränatale Bluttests. Pro und Contra' (2018) 114(44) Deutsches Ärzteblatt A1989; Freiherr von Ulmenstein, 'Tagungsbericht: Nicht-invasive Pränataldiagnostik als GKV-Leistung? – Medizinische, ethische und rechtliche Fragen' (2018) 36(9) MedR p. 680.

Considering the criteria that guide the inclusion of a new technology in the statutory health insurance,[1447] NIPT seems to be an excellent candidate to be included in the medical care that is offered during pregnancy.[1448] Compared to invasive prenatal diagnoses NIPT is not only cheaper[1449] but also safer. Amniocentesis and chorionic villus sampling, being invasive procedures, are deemed to be dangerous due to their – albeit low – potential to cause miscarriages. Some legal scholars therefore welcome the reimbursement of these tests by the health insurance,[1450] not least because it is a measure aimed at protecting the foetus from the risk of miscarriage.[1451]

By contrast, the expert opinion commissioned by the Federal Government Commissioner for Matters relating to Persons with Disabilities had claimed that reimbursement by the GKV would constitute a violation of the constitutional obligations of the state.[1452] The author had started from the assumption that Article 3(3) sentence 2 of the Basic Law requires that no one shall be discriminated against or disadvantaged because of their disability. Against this background the emergence of NIPT would trigger the state's responsibility to actively intervene to counteract the possible discrimination against people with disabilities. He argued that the early detection of a chromosomic trisomy would be likely to result in the woman's decision to undergo an abortion procedure which, depriving the foetus of the opportunity to become part of society in the first place, would

1447 According to § 135(1) SGB V, new diagnostic and treatment methods may only be provided at the expense of public health insurance funds if their diagnostic and therapeutic benefit, as well as their medical necessity and economic efficiency, are recognised and evaluated in comparison to services already included in the benefit basket, see below at sec. II.2.d.

1448 Huster, 'Der Gemeinsame Bundesausschuss als Ethikbehörde?' (2017) 35(4) MedR p. 282, 284; Huber in Steger, Orzechowski and Schochow, *Pränatalmedizin* (2018) pp. 145-146. For the clinical benefits of NIPT compared to other procedures, see Chapter 1, sec. A.I.3.b.

1449 Kießling in Rolfs and others, *BeckOK Sozialrecht* (2021) para. 11.

1450 Heinrichs, Spranger and Tambornino, 'Ethische und rechtliche Aspekte der Pränataldiagnostik' (2012) 30(10) MedR p. 625, 627; Huber in Steger, Orzechowski and Schochow, *Pränatalmedizin* (2018) pp. 146-ff; Rolfes in Steger, Orzechowski and Schochow, *Pränatalmedizin: Ethische, juristische und gesellschaftliche Aspekte* (2018) pp. 66-67.

1451 Tolmein, 'Selbstbestimmungsrecht der Frau, Pränataldiagnostik und die UN-Behindertenrechtskonvention' (2012) 45(4) KJ p. 420, 428; Kießling in Rolfs and others, *BeckOK Sozialrecht* (2021) para. 11.

1452 Gärditz, 'Gutachtliche Stellungnahme zur Zulässigkeit des Diagnostikprodukts "PraenaTest"', 2012, p. 10.

allegedly be the "most intense" form of discrimination.[1453] The outcome of these arguments is that the provision of financial support by the state for the performance of this test, such as the inclusion in the catalogue of the statutory health insurance, would constitute a breach of Article 3(3) sentence 2 of the Basic Law.[1454]

However, the opinion was silent on the invasive and more dangerous prenatal diagnostic procedures that are already offered by the statutory health insurance. As other commentators have noted from a legal perspective, the non-invasiveness of the test does not imply a qualitative leap in its potential to lead to constitutional violations.[1455] NIPT itself does not detect more disabilities, but only detects them in a less invasive way and thus with greater respect for the health and safety of the foetus.[1456] Therefore, the non-invasiveness of the test has no consequences for its legal assessment compared to the other diagnoses that are already publicly funded.[1457] From this point of view NIPT is indeed more compatible with, what part of the legal literature considers to be, the main purpose of prenatal care offered by the statutory health insurance. Namely, to avoid danger to the health and life of the mother and child.[1458]

Moreover, the expert's opinion disregarded the legal consequences of the fact that a possible abortion following NIPT is caused by the mother's decision and not by the performance of the diagnosis.[1459] Any disadvantage to the foetus would derive from the need to avoid a future risk to the health of the pregnant woman and would therefore be justified by the protection of her life and physical integrity.[1460] Rather, from the point of view of protecting the woman's physical integrity – and that of the foetus – the non-reimbursement of the least invasive test, while reimbursing more dangerous

1453 ibid, p. 4 (author's translation).
1454 ibid, p. 10.
1455 Tolmein, 'Selbstbestimmungsrecht der Frau, Pränataldiagnostik und die UN-Behindertenrechtskonvention' (2012) 45(4) KJ p. 420, 430; Kießling in Rolfs and others, *BeckOK Sozialrecht* (2021) para. 11.
1456 Kießling in Rolfs and others, *BeckOK Sozialrecht* (2021) para. 11.
1457 Heinrichs, Spranger and Tambornino, 'Ethische und rechtliche Aspekte der Pränataldiagnostik' (2012) 30(10) MedR p. 625, 629; Tolmein, 'Selbstbestimmungsrecht der Frau, Pränataldiagnostik und die UN-Behindertenrechtskonvention' (2012) 45(4) KJ p. 420, 430.
1458 Welti in Becker and Kingreen, *SGB V* (2020) para. 4.
1459 Huber in Steger, Orzechowski and Schochow, *Pränatalmedizin* (2018) p. 149.
1460 Hufen, 'Verfassungsrechtliche Bedenken gegen frühe Pränataldiagnostik?' (2017) 35(4) MedR p. 277, 281.

diagnostic methods, is problematic. Women with limited financial means would be *de facto* excluded from access to the less invasive procedure and therefore, as a consequence of their economic condition, would have to bear the risk of a miscarriage.[1461]

b Right to Know and Right Not to Know

A mother's right to know the health status of the foetus derives directly from her fundamental right to physical integrity, as set out in Article 2(1) sentence 1 of the Basic Law, which the state is obliged to protect. The right to physical integrity also includes the right to know about one's own health condition according to the current state of medical knowledge. In the case of a pregnant woman this extends to all the physical and psychological hazards that may arise from the pregnancy.[1462] The right to be informed of all conditions relevant to one's health is also supported by the fundamental right to informational self-determination that is guaranteed by Article 2(1) in conjunction with Article 1 of the Basic Law.[1463]

On the other hand, the right to physical integrity and informational self-determination equally encompass a 'right not to know',[1464] given that merely obtaining genetic information can seriously affect some patients.[1465]

1461 Heinrichs, Spranger and Tambornino, 'Ethische und rechtliche Aspekte der Pränataldiagnostik' (2012) 30(10) MedR p. 625, 628; Rolfes in Steger, Orzechowski and Schochow, *Pränatalmedizin* (2018) pp. 63-ff.

1462 Hufen, 'Zur verfassungsrechtlichen Beurteilung frühzeitiger pränataler Diagnostik', 4.1.2013, p. 22; Fündling, *Recht auf Wissen vs. Recht auf Nichtwissen in der Gendiagnostik* (2017) pp. 174-176.

1463 Fündling, *Recht auf Wissen vs. Recht auf Nichtwissen in der Gendiagnostik* (2017) pp. 149-165.

1464 See Joschko, *Das Recht auf Nichtwissen in der Gesundheitsversorgung* (2022) pp. 53-61. Particularly with regard to NIPT, criticism that it may undermine the right not to know was reported by Gärditz, 'Gutachtliche Stellungnahme zur Zulässigkeit des Diagnostikprodukts "PraenaTest"', 2012, p. 15; Hufen, 'Verfassungsrechtliche Bedenken gegen frühe Pränataldiagnostik?' (2017) 35(4) MedR p. 277, 281; Huber in Steger, Orzechowski and Schochow, *Pränatalmedizin* (2018) pp. 151-152.

1465 Fündling, *Recht auf Wissen vs. Recht auf Nichtwissen in der Gendiagnostik* (2017) p. 178; Laufs and Rehborn in Laufs, Kern and Rehborn, *Handbuch des Arztrechts* (5th edn 2019) para. 85; Kämmerer and Kunig in Münch and Kunig, *Grundgesetz* (2021) para. 80.

The 'right to know' and the 'right not to know' must equally receive sufficient protection when the patient is presented with the option to undergo prenatal screening. Adequate safeguard of both rights appears to be accomplished by the provisions of the Genetic Diagnosis Act (*Gendiagnostikgesetz,* GenDG) on informed consent and counselling.[1466] § 15 of the GenDG deals specifically with prenatal diagnosis and provides that testing may only be carried out if the pregnant woman has been duly informed, has given her consent and has received appropriate genetic counselling.[1467]

Moreover, before asking for consent, the medical practitioner responsible must inform the patient of the nature, significance and scope of the test, including the characteristics of the condition being tested for and their right not to know.[1468] Following this information the patient must be given an appropriate period of time before giving their consent.[1469] The latter must be in writing and may be revoked at any time.[1470]

Genetic counselling should be offered both before and after a prenatal genetic test.[1471] Counselling takes place at a separate time from the provision of information and the taking of informed consent and forms part of the treatment itself.[1472] Counselling must be 'non-directive', meaning that it must be impartial and should aim at assisting the patient in forming their own opinion.[1473]

1466 Fündling, *Recht auf Wissen vs. Recht auf Nichtwissen in der Gendiagnostik* (2017) p. 313.

1467 On this point see Joerden and Uhlig in Steger, Ehm and Tchirikov, *Pränatale Diagnostik und Therapie in Ethik, Medizin und Recht* (2014) pp. 105-107.

1468 § 9(2) no. 5 GenDG, see Fündling, *Recht auf Wissen vs. Recht auf Nichtwissen in der Gendiagnostik* (2017) p. 312.

1469 § 9 GenDG.

1470 § 8 GenDG.

1471 § 15 GenDG. On the differences between counselling before and after the testing, see Joerden and Uhlig in Steger, Ehm and Tchirikov, *Pränatale Diagnostik und Therapie in Ethik, Medizin und Recht* (2014) p. 107.

1472 Fündling, *Recht auf Wissen vs. Recht auf Nichtwissen in der Gendiagnostik* (2017) p. 225.

1473 Fenger in Spickhoff, *Medizinrecht* (3rd edn 2018) para. 3; Laufs and Rehborn in Laufs, Kern and Rehborn, *Handbuch des Arztrechts* (2019) para. 84.

2. The G-BA's Assessment of NIPT

a Reactions to the Initiation of the Procedure

Right after it was placed on the market the price of NIPT was very significant and represented a major financial obstacle for most patients.[1474] In 2013 the manufacturer submitted an application to the G-BA to initiate a medical device evaluation procedure under § 137e SGB V. According to this provision the Federal Joint Committee can evaluate new medical devices through a 'coverage with evidence development' procedure, which could also be initiated upon application of the manufacturer.[1475] This procedure allows for the temporary reimbursement,[1476] in a trial stage,[1477] of a medical device or medical treatment whose benefits have not yet been sufficiently proven.[1478]

The G-BA's announcement that a consultative procedure was launched, leading to a 'coverage with evidence development' procedure for NIPT,[1479] revived the heated ethical debate.[1480]

In particular, an article published in January 2015 by the weekly *Zeit* denounced the prospective reimbursement of NIPT by the statutory health insurance as the first step towards a society that wants to get rid of people with congenital disabilities.[1481] The G-BA was forced to respond to these allegations by publishing an official position stating its awareness of the

1474 Schmitz, 'Ethische Herausforderungen der neuen nichtinvasiven Pränataltestung' (2016) 49(6) Gynäkologe p. 442, 443; Braun and Könninger, 'Realizing Responsibility.' (2017) 37(3) New Genetics and Society p. 248, 251.

1475 According to § 137e (7) SGB V.

1476 According to § 137e(1) sentence 2 SGB V.

1477 The trial period is meant to collect additional data that will be used to reach the final decision on the reimbursement of the device or procedure by the statutory health insurance. On the data collection, see Becker in Becker and Kingreen, *SGB V: Gesetzliche Krankenversicherung Kommentar* (7th edn 2020) para. 9.

1478 Becker in Becker and Kingreen, *SGB V* (2020) para. 3; Propp in Rolfs and others, *BeckOK Sozialrecht* (61st edn 2021) para. 5–6.

1479 Gemeinsamer Bundesausschuss (G-BA), 'Pressemitteilung Nr. 20/2014: Methodenbewertung: Erprobung von neuen Untersuchungs- und Behandlungsmethoden: Weiterer Meilenstein erreicht' (8.5.2014) <https://www.g-ba.de/downloads/34-215-534/20-2014-05-08_Erprobungsrichtlinien.pdf> accessed 15.12.2019.

1480 As reported by Braun and Könninger, 'Realizing Responsibility.' (2017) 37(3) New Genetics and Society p. 248, 260; Huster, 'Der Gemeinsame Bundesausschuss als Ethikbehörde?' (2017) 35(4) MedR p. 282, p. 284.

1481 Bahnsen, 'Pränataldiagnostik: Der Test' *Die Zeit* (22.1.2015) <https://www.zeit.de/2015/04/praenataldiagnostik-down-syndrom-krankenkasse> accessed 28.9.2021.

ethical concerns surrounding NIPT.[1482] On this occasion the chairman of the Federal Joint Committee clarified that the trial procedure and reimbursement of costs for study participants had not yet begun and reassured the public that the committee would treat ethical issues with great sensitivity.

A similar statement was issued the following year, on the occasion of the G-BA's decision to discontinue the trial procedure in order to start a regular assessment of the medical device as per § 135(1) SGB V.[1483] As the collection of further data to assess the benefits of NIPT was not found to be necessary for its evaluation,[1484] a regular assessment procedure, aimed at obtaining a definitive inclusion of NIPT in the statutory health insurance, had been initiated on 4 July 2016 by an application of the National Association of Statutory Health Insurance Funds, the National Association of Statutory Health Insurance Physicians as well as the chairman and impartial members of the G-BA.[1485]

The announcement of the application to start the regular procedure triggered a reaction from a group of MPs who, in a letter to the G-BA, claimed that the test did not offer any medical benefit.[1486] The letter urged the G-BA

1482 Gemeinsamer Bundesausschuss (G-BA), 'Pressemitteilung Nr. 02/2015: Methodenbewertung: Klarstellung des Sachstandes zu Pränatests für Schwangere' (22.1.2015) <https://www.g-ba.de/downloads/34-215-566/02-2015-01-22_Erprobung.pdf> accessed 10.8.2022.

1483 Gemeinsamer Bundesausschuss (G-BA), 'Pressemitteilung Nr. 32/ 2016: Methodenbewertung: Nicht-invasive Pränataldiagnostik bei Risiko-schwangerschaften - G-BA beginnt Verfahren zur Methodenbewertung - Beratungen zur Erprobung ruhend gestellt' (18.8.2016) <https://www.g-ba.de/downloads/34-215-635/32_2016-08-18_Methodenbewertung%20NIPD.pdf> accessed 28.9.2021.

1484 Huster, 'Der Gemeinsame Bundesausschuss als Ethikbehörde?' (2017) 35(4) MedR p. 282, 284; Richter-Kuhlmann, 'Nicht invasive Pränataldiagnostik: Es geht um mehr als nur Geld' (2019) 116(16) Deutsches Ärzteblatt A774-A778, A778.

1485 Gemeinsamer Bundesausschuss (G-BA), 'Antrag auf Bewertung der Methode der nicht-invasiven Pränataldiagnostik (NIPD) zur Bestimmung des Risikos autosomaler Trisomien 13, 18 und 21 mittels eines molekular-genetischen Tests für die Anwendung bei Risikoschwangerschaften im Rahmen der Mutterschafts-Richtlinien nach § 135 Absatz 1 SGB V' (4.7.2016) <https://www.g-ba.de/downloads/40-268-3933/2016-08-18_Einleitung-Beratungsverf_nicht-invasive-Praenataldiagnostik_Antrag.pdf> accessed 28.9.2021.

1486 Hüppe and others, 'TOP 8.2.1 der 91. Öffentlichen G-BA Sitzung am 18. August 2016' (17.8.2016) <https://www.netzwerk-praenataldiagnostik.de/data/praenatal-diagnostik/pdf/Brief_MdBs_zur_91_G-BA-Sitzung.pdf> accessed 28.9.2021. See Deutscher Bundestag, 'BT-Drucks. 19/9059: Bericht des Ausschusses für Bildung, Forschung und Technikfolgenabschätzung (18. Ausschuss) gemäß § 56a der

to consider ethical and social consequences in the medical evaluation of the test and to involve associations of people with disabilities in the procedure.[1487] A further letter, coming from a network against selection through prenatal diagnosis and other stakeholders, expressed similar concerns.[1488]

In response, in the statement accompanying the press release that was issued upon the launch of the procedure, the chairman of the committee reiterated that the ethical concerns raised by NIPT would be taken into account and that the German Ethics Council and other social or scientific organisations would be consulted during the procedure.[1489]

b Health Technology Assessment

The authority responsible for health technology assessment in Germany,[1490] namely the Institute for Quality and Efficiency in Health Care (*Institut für Qualität und Wirtschaftlichkeit im Gesundheitswesen*, IQWiG), performs a preparatory function for the G-BA's issuing of guidelines.[1491] The Institute's tasks include the research, presentation and evaluation of the current state of medical knowledge on diagnostic and therapeutic procedures, as laid down in § 139a(3) no. 1 of the SGB V.

As a first step in the evaluation procedure of NIPT the G-BA decided to commission an assessment by the IQWiG of the current state of medical knowledge on NIPT, with a view to its possible use in high-risk pregnancies within the framework of the maternity guidelines.[1492] Furthermore, the HTA authority was instructed to prepare an informative brochure for

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Geschäftsordnung' (4.4.2019), p. 68. <https://dserver.bundestag.de/btd/19/090/1909059.pdf> accessed 28.9.2021.

1487 Hüppe and others, 'TOP 8.2.1 der 91. Öffentlichen G-BA Sitzung am 18. August 2016', 17.8.2016; Andorno, 'The Precautionary Principle' (2004) 1(1) JIBL p. 11.

1488 See Deutscher Bundestag, 'BT-Drucks. 19/9059', 4.4.2019, p. 67.

1489 Gemeinsamer Bundesausschuss (G-BA), 'Pressemitteilung Nr. 32/ 2016', 18.8.2016.

1490 On the IQWiG as the German authority for HTA, see Widrig, *Health Technology Assessment* (2015) pp. 348-ff.

1491 Wallrabenstein in Becker and Kingreen, *SGB V: Gesetzliche Krankenversicherung Kommentar* (7th edn 2020) para. 1.

1492 Gemeinsamer Bundesausschuss (G-BA), 'Konkretisierung des Auftrags des Gemeinsamen Bundesausschusses an das Institut für Qualität und Wirtschaftlichkeit im Gesundheitswesen: Evidenzbewertung der nicht-invasiven Pränataldiagnostik (NIPD) zur Bestimmung des Risikos autosomaler Trisomien 13, 18 und 21 mittels eines molekulargenetischen Tests für die Anwendung bei Risikoschwangerschaften im Rahmen der Mutterschafts-Richtlinien (Mu-RL)'

insured persons on the existing options for prenatal diagnosis under the Maternity Guidelines.[1493]

In a preliminary report the IQWiG declared that an assessment of the scientific studies indicated that NIPT was very accurate and reliable for trisomy 21.[1494] While for trisomies 13 and 18 the results were less conclusive, NIPT showed potential to significantly reduce the number of possible miscarriages due to invasive diagnoses.[1495] Based on these considerations the IQWiG evaluated several scenarios for the possible integration of NIPT into the prenatal care pathway offered by the Maternity Guidelines.[1496] Its assessment was published in a preliminary draft and open to comments from all interested individuals, institutions and organisations.[1497]

After the public consultation phase, the final assessment was published in June 2018.[1498] In response to comments criticising the lack of consideration of ethical issues, a paragraph on ethical dimensions was added to the final report. However, the section only stated that the ethical dimension of NIPT was known to the G-BA as a final decision-making body and should therefore not be addressed in the health technology assessment.[1499]

(26.1.2017) <https://www.g-ba.de/downloads/40-268-4204/2017-01-26_Mu-RL_Auftragskonkretisierung_Evidenzbewertung.pdf> accessed 28.9.2021.

1493 Gemeinsamer Bundesausschuss (G-BA), 'Beschluss über eine Beauftragung des Instituts für Qualität und Wirtschaftlichkeit im Gesundheitswesen: Erstellung einer Versicherteninformation über die bestehenden Möglichkeiten der Pränataldiagnostik gemäß Mutterschafts-Richtlinien (Mu-RL) sowie der Einbindung von Eckpunkten, die sich gegebenenfalls aus einer zukünftigen Änderung der Mu-RL ergeben' (16.2.2017) <https://www.g-ba.de/downloads/39-261-2857/2017-02-16_Mu-RL_IQWiG-Beauftragung-Versicherteninformation-PD-NIPD.pdf> accessed 28.9.2021.

1494 Institut für Qualität und Wirtschaftlichkeit im Gesundheitswesen, 'Nicht invasive Pränataldiagnostik (NIPD) zur Bestimmung des Risikos autosomaler Trisomien 13, 18 und 21 bei Risikoschwangerschaften (Vorbericht)' (11.12.2017), p. 21 <https://www.iqwig.de/download/s16-06_nicht-invasive-praenataldiagnostik-nipd_vorbericht_v1-0.pdf?rev=187029> accessed 28.9.2021.

1495 ibid.

1496 ibid, p. 71.

1497 ibid, p. III.

1498 Institut für Qualität und Wirtschaftlichkeit im Gesundheitswesen, 'Nicht invasive Pränataldiagnostik kann Trisomie 21 zuverlässig bestimmen' (27.6.2018) <https://www.iqwig.de/presse/pressemitteilungen/pressemitteilungen-detailseite_10172.html> accessed 28.9.2021.

1499 Institut für Qualität und Wirtschaftlichkeit im Gesundheitswesen, 'IQWiG-Berichte - Nr. 623: Nicht invasive Pränataldiagnostik (NIPD) zur Bestimmung des Risikos autosomaler Trisomien 13, 18 und 21 bei Risikoschwangerschaften (Ab-

The IQWiG's refusal to address possible ethical problems has been criticised as a failure to reflect on the qualification of fetal disability as a problem to be diagnosed[1500] and consequently on the existence of a medical benefit in the use of the test.[1501]

c Consultation and Parliamentary Debate

Based on the health technology assessment report the G-BA published a draft decision in March 2019. The key points of the draft were that NIPT should only be reimbursed by the statutory health insurance with a view to the individual circumstances of the pregnant woman and after the 12th week of pregnancy.[1502] A purely statistical risk due to the mother's age would therefore not be sufficient to qualify for reimbursement. The aim of including NIPT in prenatal care would be to enable the pregnant woman to face the possible presence of a trisomy while avoiding invasive diagnoses that could lead to miscarriages. To achieve this the draft envisaged that the mother would be provided with comprehensive counselling and information.[1503]

Upon publication of the draft the G-BA initiated a formal consultation procedure. In accordance with §§ 91(5) and 91(5a), §§ 92(1b) and 92(7d) of the SGB V written comments were solicited from the German Medical Association, the Federal Commissioner for Data Protection and Freedom

schlussbericht)' (30.4.2018), p. 85 <https://www.iqwig.de/download/s16-06_nicht-invasive-praenataldiagnostik-nipd_abschlussbericht_v1-0.pdf> accessed 28.9.2021.

1500 Deutscher Bundestag, 'BT-Drucks. 19/9059', 4.4.2019, p. 67.

1501 Braun and Könninger, 'Realizing Responsibility.' (2017) 37(3) New Genetics and Society p. 248, 260; BioSkop, GeN and Netzwerk gegen Selektion durch Pränataldiagnostik, 'Gemeinsame Stellungnahme zum Bericht der IQWiG: "Nicht invasive Pränataldiagnostik zur Bestimmung des Risikos autosomaler Trisomien 13, 18 und 21 bei Risikoschwangerschaften": Moratorium für den Bluttest!' (4.7.2018) <https://gen-ethisches-netzwerk.de/sites/default/files/dokumente/2018-07/2018_07_04-stellungnahme-gen_iqwig.pdf> accessed 28.9.2021.

1502 Gemeinsamer Bundesausschuss (G-BA), 'Tragende Gründe zum Beschlussentwurf über eine Änderung der Mutterschafts-Richtlinien (Mu-RL): Nicht-invasive Pränataldiagnostik zur Bestimmung des Risikos autosomaler Trisomien 13, 18 und 21 mittels eines molekulargenetischen Tests (NIPT) für die Anwendung bei Risikoschwangerschaften' (22.3.2019), pp. 3-ff <https://www.g-ba.de/downloads/40-268-5640/2019-03-22_Einleitung-SN_NiPT_Beschlussentwurf_TrG_WZ.pdf> accessed 29.9.2021.

1503 ibid, p. 4.

of Information, midwives associations, professional societies, organisations of medical device manufacturers and NIPT manufacturers. [1504] At the same time, as part of a wider public debate, the G-BA also called on the German Ethics Council and the Genetic Diagnostics Commission (*Gendiagnostik-Kommission*, GEKO) to comment on the draft,[1505] and on the Bundestag to initiate a parliamentary debate on the political and normative aspects.[1506]

The German Ethics Council declined to intervene, but referred back to the statement it had already issued in 2013 on the future of genetic diagnostics.[1507] On that occasion the Council had stated that NIPT serves medical purposes and could therefore be offered by the statutory health insurance in the case of pregnancies at increased risk. A dissenting opinion signed by four members had on the contrary argued that NIPT should not be supported by public funding and should not be part of the services offered by the statutory health insurance.[1508]

As for the Bundestag, a parliamentary 'orientation debate' on the issue of NIPT reimbursement by the GKV was conducted in April 2019.[1509]

Whereas no MPs were in favour of a routine screening of trisomies that would be provided indiscriminately to all pregnant women, most agreed that NIPT should be offered by statutory the health insurance instead of the riskier invasive diagnoses already carried out.[1510] In addition, some speakers pointed out that integrating NIPT into the Maternity Guidelines

1504 Gemeinsamer Bundesausschuss (G-BA), 'Beschluss des Gemeinsamen Bundesausschusses über die Einleitung des Stellungnahmeverfahrens gemäß § 91 Absatz 5, § 91 Absatz 5a sowie § 92 Absatz 1b und § 92 Absatz 7d des Fünften Buches Sozialgesetzbuch (SGB V) vor einer abschließenden Entscheidung über eine Änderung der Mutterschafts-Richtlinien: Nicht-invasive Pränataldiagnostik (NIPD) autosomaler Trisomien 13, 18 und 21 mittels eines molekulargenetischen Tests (NIPT) für die Anwendung bei Risikoschwangerschaften im Rahmen der Mutterschafts-Richtlinien (Mu-RL)' (22.3.2019).

1505 Gemeinsamer Bundesausschuss (G-BA), 'Nicht-invasive Tests bei Risikoschwangerschaften: G-BA fordert zur Stellungnahme auf' (22.3.2019) <https://www.g-ba.de/presse/pressemitteilungen-meldungen/789/> accessed 28.9.2021.

1506 Braun and Könninger, 'Realizing Responsibility.' (2017) 37(3) New Genetics and Society p. 248, 262.

1507 As reported by Deutscher Bundestag, 'BT-Drucks. 19/9059', 4.4.2019, p. 67.

1508 Deutscher Ethikrat, 'The Future of Genetic Diagnosis' (2013) p. 167.

1509 Deutscher Bundestag, 'Plenarprotokoll 19/95: 95. Sitzung' (Berlin 11.4.2019), pp. 11315-ff.

1510 Deutscher Bundestag, 'Plenarprotokoll 19/95: 95. Sitzung' (Berlin 11.4.2019), see *inter alia*, the speeches of Karl Lauterbach; Cornelia Möhring, Volker Münz, Katja Dörner, Katrin Helling-Plahr, Stephan Pilsinger.

would guarantee adequate information counselling for pregnant women, which could otherwise not be ensured in the private sector.[1511] Moreover, the fact that the tests would still be available out-of-pocket to those women who have sufficient financial resources was considered by many MPs to be discriminatory. Women with fewer financial means would be forced to accept a certain risk of miscarriage in order to obtain information on the health of the foetus.[1512] As one speaker put it: the reimbursement of costs by statutory health insurance can be seen as not an ethical but rather a social issue.[1513]

In opposition to this, a consistent minority of MPs argued against the public funding of NIPT. Some claimed that it would be incompatible with the purpose of the public healthcare system to treat individuals[1514] and that the state should not actively bring about the conditions for the abortion of foetuses with chromosomal trisomies, as such a value choice would be ethically and politically wrong.[1515] Others emphasised that the possibility for a woman to decide free of pressure and her right not to know, as well as the importance of an inclusive society, were essential values that were at stake.[1516]

However, this parliamentary debate has not been followed up upon to date. As the scheduled date for the G-BA's final decision on the amendments to the Maternity Guidelines approached, a group of MPs decided to address the G-BA members directly. They sent a letter asking them to consider suspending the procedure and the decision in order not to prevent further parliamentary discussions.[1517]

1511 Deutscher Bundestag, 'Plenarprotokoll 19/95: 95. Sitzung' (Berlin 11.4.2019), see the interventions of Claudia Schmidtke and Thomas Rachel.

1512 Deutscher Bundestag, 'Plenarprotokoll 19/95: 95. Sitzung' (Berlin 11.4.2019), *inter alia,* Karl Lauterbach, Christine Aschenberg-Dugnus, Petra Sitte, Katrin Helling-Plahr, Marja-Liisa Völlers.

1513 Deutscher Bundestag, 'Plenarprotokoll 19/95: 95. Sitzung' (Berlin 11.4.2019), p. 11337, intervention by Erwin Rüddel.

1514 Deutscher Bundestag, 'Plenarprotokoll 19/95: 95. Sitzung' (Berlin 11.4.2019), p. 11319, Corinna Rüffer.

1515 Matthias Bartke in Deutscher Bundestag, 'Plenarprotokoll 19/95: 95. Sitzung' (Berlin 11.4.2019)

1516 Dagmar Schmidt and Christine Aschenberg-Dugnus in Deutscher Bundestag, 'Plenarprotokoll 19/95: 95. Sitzung' (Berlin 11.4.2019), pp. 11318-11321.

1517 As explained in the G-BA chairman's reply to the letter of the MPs, Gemeinsamer Bundesausschuss (G-BA), 'Schreiben von Prof. Josef Hecken, unparteiischer Vorsitzender des G-BA, an Mitglieder des Deutschen Bundestages zur Nichtvertagung der Beschlussfassung zu NIPT' (19.9.2019) <https://www.g-ba.de/

The response letter from the chairman of the G-BA stated that the committee had unanimously decided to continue the procedure. The inclusion of NIPT in the maternity guidelines would serve primarily to avoid high-risk invasive diagnoses. The chairman acknowledged that the committee was aware of the fundamental ethical issues at stake but argued that these require a legislative response. For these purposes the letter pointed out that the committee had left room for discussion and possible parliamentary decision on the issue during the three years of the assessment procedure. Moreover, the decision on the amendments to the maternity guidelines would not, in any case, preclude other initiatives by Parliament, which remained free to intervene with a legislative act to revise the G-BA decision. Lastly, the chairman mentioned that the G-BA decision would not yet warrant any claim for reimbursement. An assumption of costs by the GKV could not take place until the information brochure for insured persons was adopted.[1518]

Beside the feedback from the Bundestag, the G-BA had received a total of 30 comments from the other associations called upon to intervene. These were taken into account when reformulating the final decision.[1519]

d Inclusion of NIPT in the Maternity Guidelines

In its final decision of 19 September 2019 the G-BA amended the Maternity Guidelines to include NIPT in the prenatal care pathway. In order to avoid invasive diagnostic measures reimbursement of NIPT by the statutory health insurance was foreseen in those cases where "it is necessary to enable a pregnant woman to discuss her individual situation with regard to the presence of a trisomy within the framework of medical support".[1520] It was

downloads/17-98-4847/2019-09-19-PA-JHecken_an-BT-Abgesordnete_NIPT.pdf> accessed 28.9.2021.

1518 ibid.

1519 Gemeinsamer Bundesausschuss (G-BA), 'Pressemitteilung Nr. 26/2019: Nicht-invasiver Test zum Vorliegen von Trisomien als mögliche Alternative zu invasivem Eingriff' (19.9.2019) <https://www.g-ba.de/downloads/34-215-810/26_2019-09-19_Mu-RL_NIPT.pdf> accessed 28.9.2021.

1520 Gemeinsamer Bundesausschuss (G-BA), 'Beschluss über eine Änderung der Mutterschafts-Richtlinien (Mu-RL): Nicht-invasive Pränataldiagnostik zur Bestimmung des Risikos autosomaler Trisomien 13, 18 und 21 mittels eines molekulargenetischen Tests (NIPT) für die Anwendung bei Schwangerschaften mit besonderen Risiken' (19.9.2019) BAnz AT 20.12.2019 B6, p. 3 <https://www.g-ba

again pointed out that a statistically increased risk of trisomy would not be sufficient to access this test.[1521]

In explaining the reasons for the decision the Committee addressed the three criteria regulating the inclusion of new diagnostic or therapeutic services in the benefit basket of the GKV according to § 135(1) no. 1 SGB V. Namely, diagnostic or therapeutic benefit, medical necessity and economic efficiency. The provision also states that these aspects must be evaluated in comparison with other services already included in the benefit basket.

The diagnostic benefit and medical necessity of NIPT were determined, on the one hand, on the basis of the possibility it offered to replace invasive diagnoses and lower the risk of miscarriages and, on the other hand, on the grounds of its high specificity and sensitivity, which reduces the amount of false positives and false negatives.[1522] More generally, the medical necessity of prenatal diagnosis was grounded on the need to enable pregnant women to confront the possibility of fetal trisomies and to assess, within a medical framework, whether the pregnancy could result in a serious impairment of the physical or psychical health of the patient.[1523]

As regards the criterion of economic efficiency, the G-BA acknowledged that the reimbursement of NIPT would lead to additional costs for the public healthcare system. However, economic efficiency would be ensured by decreasing costs for the avoidable invasive diagnoses and related complications.[1524]

As part of the measures to ensure quality of care the final decision included comprehensive counselling and information for the pregnant woman. The requirements that such information must fulfil were based on the provisions contained in the Genetic Diagnosis Act. Counselling must therefore include a thorough discussion of possible medical, psychological and social issues related to the test and the consequences of the results.

.de/downloads/39-261-3955/2019-09-19_Mu-RL_NIPT_BAnz_WZ.pdf> accessed 28.9.2021 (author's translation).

1521 ibid.

1522 Gemeinsamer Bundesausschuss (G-BA), 'Tragende Gründe zum Beschluss über eine Änderung der Mutterschafts-Richtlinien (Mu-RL): Nicht-invasive Pränataldiagnostik zur Bestimmung des Risikos autosomaler Trisomien 13, 18 und 21 mittels eines molekulargenetischen Tests (NIPT) für die Anwendung bei Schwangerschaften mit besonderen Risiken' (19.9.2019), p. 3 <https://www.g-ba.de/downloads/40-268-6007/2019-09-19_Mu-RL_NIPT_TrG.pdf> accessed 28.9.2021.

1523 ibid, p. 4.

1524 ibid, p. 7.

After counselling, the woman must be given a reasonable period of time to reflect before the test.[1525] The right not to know must also be guaranteed at all stages of the procedure in accordance with the Genetic Diagnosis Act.[1526] It is prescribed that the information shall be given on the basis of an informative brochure for insured persons. As stated in the Press Release accompanying the decision, the amendments to the Maternity Guidelines concerning the use of NIPT would only come into force with the approval of this informative brochure. Until then, G-BA decision did not ground any right to claim a reimbursement of NIPT by the GKV.[1527]

After another consultation procedure in which comments were again solicited from the German Ethics Council and the GEKO – among other organisations –[1528] the information for insured subjects was approved as an annex to the maternity guidelines in August 2021.[1529] The information

1525 ibid, p. 5.

1526 ibid. On the counselling and informed consent requirement provided for by the Genetic Diagnosis Act, see above in this section at para. II.1.b. Initially, there was some doubt as to whether the provisions of the Genetic Diagnosis Act could also be applied to NIPT, see *inter alia* Lindner, 'Fällt der "PraenaTest" in den Anwendungsbereich des §15 GenDG?' (2013) 31(5) MedR p. 288. However, doubts were soon removed thanks to a statement by the Commission on Genetic Testing (*Gendiagnostik-Kommission, GEKO*), '8. Mitteilung der GEKO zur Einordnung der nicht-invasiven Pränataldiagnostik (NIPD) und der diesbezüglichen Beratungsqualifikation' (12.3.2014) <https://www.rki.de/DE/Content/Kommissionen/GendiagnostikKommission/Mitteilungen/GEKO_Mitteilungen_08.html> accessed 13.4.2022, see Hübner and Pühler in Katzenmeier and Ratzel, *Festschrift für Franz-Josef Dahm* (2017) pp. 257-258.

1527 Gemeinsamer Bundesausschuss (G-BA), 'Pressemitteilung Nr. 26/2019', 19.9.2019.

1528 Gemeinsamer Bundesausschuss (G-BA), 'Beschluss zur Einleitung des Stellungnahmeverfahrens gemäß § 91 Absatz 5, § 92 Absatz 1b und § 92 Absatz 7d des Fünften Buches Sozialgesetzbuch (SGB V) sowie gemäß 1. Kapitel § 8 Absatz 2 Satz 1 lit. a) VerfO vor einer abschließenden Entscheidung über eine Änderung der Mutterschafts-Richtlinien: Aufnahme einer Versicherteninformation zur Nicht-invasiven Pränataldiagnostik zur Bestimmung des Risikos autosomaler Trisomien 13, 18 und 21 mittels eines molekular-genetischen Tests (NIPT-Trisomie 13,18,21) für die Anwendung bei Schwangerschaften mit besonderen Risiken' (22.4.2021) <https://www.g-ba.de/downloads/39-261-4803/2021-04-22_Mu-RL_Einleitung-SN-Versicherteninfo-NIPT.pdf> accessed 28.9.2021.

1529 Gemeinsamer Bundesausschuss (G-BA), 'Beschluss über eine Änderung der Mutterschafts-Richtlinien (Mu-RL): Aufnahme einer Versicherteninformation zur Durchführung der Nicht-invasiven Pränataldiagnostik zur Bestimmung des Risikos autosomaler Trisomien 13, 18 und 21 mittels eines molekulargenetischen Tests (NIPT-Trisomie 13,18,21) für die Anwendung bei Schwangerschaften mit besonderen Risiken' (19.8.2021) <https://www.g-ba.de/downloads/39-261-4987/2021-08-19_Mu-RL_NIPT_Versicherteninformation.pdf> accessed 29.9.2021.

leaflet specifies that NIPT is not a generally recommended screening test and that it can only be reimbursed by statutory health insurance in exceptional cases on the basis of the specific situation of the individual patient.[1530] It is made clear that NIPT for chromosomal trisomies is not a form of routine screening and that the costs can only be covered when a woman and her doctor conclude that the test is necessary in view of the woman's personal situation, for instance in cases where uncertainty about the presence of a chromosomal trisomy in the foetus affects the woman intolerably.[1531] Alternatively, NIPT is also covered if a previous screening has already shown an increased risk of trisomies.[1532]

However, some further steps were necessary after the approval of the information brochure in order to allow patients' use of NIPT at the expenses of statutory health insurance funds. Firstly, as required by § 94 SGB V, the Federal Ministry of Health was given two months to submit a possible objection to the inclusion of the informative brochure in the maternity guidelines.[1533] Following this, the guidelines containing the annex could be published in the Federal Gazette (*Bundesanzeiger*).[1534] Finally, the health insurance funds and the doctors' representatives were to negotiate the details regarding the invoicing of the test and the counselling service within six months.[1535] On the 18 May 2022 the evaluation committee (*Bewertungsausschuss*) in charge of this decision agreed on the details of the reimbursement of NIPT for the determination of the risk of trisomies 13, 18 and 21, including the medical consultation prior to the test.[1536] Based on

1530 Gemeinsamer Bundesausschuss (G-BA), Mutterschafts-Richtlinien 10.12.1985, p. 44.

1531 ibid p. 48.

1532 ibid.

1533 The approval of the Federal Ministry of Health was also required for the adoption of the previous changes to the maternity guidelines, on that occasion it promptly arrived at the end of November 2019, see document available at <https://www.g-ba.de/downloads/40-268-6166/2019-09-19_Mu-RL_NIPT_BMG.pdf> accessed 28.9.2021.

1534 According to § 94(2) SGB V.

1535 Gemeinsamer Bundesausschuss (G-BA), 'Pressemitteilung Nr. 28/2021: Versicherteninformation zum vorgeburtlichen Bluttest auf Trisomien liegt nun vor' (19.8.2021) <https://www.g-ba.de/presse/pressemitteilungen-meldungen/974/> accessed 29.8.2021.

1536 Kassenärztliche Bundesvereinigung, 'Beschluss des Bewertungsausschusses nach § 87 Abs. 1 Satz 1 SGB V in seiner 594. Sitzung am 18. Mai 2022 zur Änderung des Einheitlichen Bewertungsmaßstabes (EBM)' (2022) 119(24) Deutsches Ärzteblatt A1108-A1111.

this decision NIPT has been reimbursed by the statutory health insurance funds – in the individual cases provided for by the maternity guidelines – starting from the 1 July 2022, i.e. six years after the start of the regular assessment procedure.

The final decision of the G-BA did not put an end to the public debate. In February 2022 the German Ethics Council held an online public discussion in view of the upcoming reimbursement of NIPT by statutory health insurance funds.[1537] Among the topics addressed were: the arguments for and against the use of NIPT, the design of appropriate counselling and the possible social consequences of NIPT routinisation.[1538] The audience was given the opportunity to participate in the panel discussion by asking questions online.

In July 2022 a group of parliamentarians again called for legislative intervention on NIPT. They expressed fear of routinisation of the test and argued that the ethically controversial decision on whether or not to reimburse NIPT under the statutory health insurance should be made by the legislature rather than the health administration.[1539]

3. *Room for Ethical Considerations in the G-BA's Assessment*

The description of the assessment procedure for NIPT shows how the G-BA decided to concentrate exclusively on the medical and scientific appraisal of the innovative prenatal diagnostic technique, while shifting responsibility for the ethical and normative aspects to other bodies such as the Bundestag and the German Ethics Council, which were called upon to intervene.[1540]

The decision to provide for the reimbursement of NIPT only after a careful assessment of the woman's personal circumstances was welcomed

1537 Deutscher Ethikrat, 'Pressemitteilung 01/2022: Ethikrat lädt ein zum Thema „Wissens-Wert? Zum verantwortlichen Umgang mit nichtinvasiven Pränataltests (NIPT)"' <https://www.ethikrat.org/mitteilungen/mitteilungen/2022/ethikrat-laedt-ein-zum-thema-wissens-wert-zum-verantwortlichen-umgang-mit-nichtinvasiven-praenataltests-nipt/?cookieLevel=not-set> accessed 6.4.2022.

1538 ibid.

1539 'Pränatale Diagnostik:"Wir stehen erst am Beginn einer besorgniserregenden Entwicklung"' *Süddeutsche Zeitung* (28.7.2022) <https://www.sueddeutsche.de/politik/praenatale-diagnostik-bundestag-trisomie-1.5629581> accessed 3.8.2022.

1540 See Braun and Könninger, 'Realizing Responsibility.' (2017) 37(3) New Genetics and Society p. 248, 262-ff.

as a pragmatic solution suited to the German context and debate.[1541] The emphasis on the individual case was said to have a legitimising effect on the G-BA's solution, which succeeded in both ensuring that NIPT does not become a routinely performed test and at the same time provides all women who deem it necessary with affordable access to the test.[1542]

However, some authors have argued that the G-BA did take an ethical stance in deciding to consider NIPT to be medically necessary.[1543] Criticism of the G-BA's decision has brought into question the very concept of fetal trisomies as medical problems requiring a diagnosis.[1544] These arguments appear unconvincing, however, when one considers that other prenatal diagnoses for detecting trisomies are reimbursed by statutory health insurance, even if they are more dangerous to the health of the foetus.[1545] Against this background, the decision to consider NIPT to be medically necessary cannot be considered an ethical choice.

On the other hand, many have argued that the G-BA should have taken into account the ethical aspects of NIPT and, based on them, decided to either suspend the assessment procedure or exclude reimbursement by the GKV.[1546]

However, these options are not compatible with the legal framework regulating the G-BA and its competences. Firstly, the G-BA cannot legitimately suspend the procedure because of ethical issues. As regards the

1541 Rehmann-Sutter and Schües, 'Die NIPT-Entscheidung des G-BA. Eine ethische Analyse' (2020) 32(4) Ethik Med p. 385, 399-400.

1542 ibid, p. 399.

1543 Braun and Könninger, 'Realizing Responsibility.' (2017) 37(3) New Genetics and Society p. 248, 262.

1544 As reported by the Bundestag report on prenatal diagnosis, Deutscher Bundestag, 'BT-Drucks. 19/9059', 4.4.2019, p. 67: The 'technicist tunnel vision of the study design' which 'unreflectively presupposes the disability of the foetus as a problem to be diagnosed' met with public criticism (author's translation). See also Freiherr von Ulmenstein, 'Tagungsbericht' (2018) 36(9) MedR p. 680, 680–681.

1545 And as Huster notes (in Huster, 'Non-invasive Prenatal Diagnostics (NIPD) in the System of Medical Care: Ethical and Legal issues' (2021) 49(8) J Perinat Med p. 1, 5), no one in the discussion suggested removing them from the benefit basket of the statutory health insurance.

1546 As was demanded of the G-BA in a letter from ten MPs, see Gemeinsamer Bundesausschuss (G-BA), 'Schreiben von Prof. Josef Hecken, unparteiischer Vorsitzender des G-BA, an Mitglieder des Deutschen Bundestages zur Nichtvertagung der Beschlussfassung zu NIPT', 19.9.2019.

coverage with evidence development procedure,[1547] the G-BA is given a deadline of three months to decide on the application of the producer.[1548] The suspension or stalling of a regular evaluation procedure could lead to a so-called 'system failure' according to § 13 SGB V if it occurs due to arbitrary reasons.[1549] Thus, in the case of a product that meets all the requirements for inclusion in the statutory health insurance, a suspension of the assessment on purely ethical grounds could have entitled patients to obtain reimbursement of NIPT directly from the public health insurance funds.[1550]

Once the evaluation procedure has started the list of aspects that have to be taken into account by the G-BA under § 135(1) of the SGB V is exhaustive.[1551] There is no legal basis that would allow the G-BA to bring ethical aspects into consideration when deciding on reimbursement by the statutory health insurance. For this reason objections had already been raised in response to the G-BA's press release that sought to reassure stakeholders and the public that the German Ethics Council would be involved in the procedure.[1552] The adoption of further evaluation criteria by the G-BA could only be made legitimate by a legal provision that integrated them into the exhaustive list in § 135(1) of the SGB V.[1553] Such a legal basis would be necessary also considering the relevance of the G-BA's guidelines for the fundamental rights of the individual.[1554]

1547 As implemented by the Act on the Improvement of Care Structures in Statutory Health Insurance (Gesetz zur Verbesserung der Versorgungsstrukturen in der gesetzlichen Krankenversicherung, GKV-VStG) which introduced § 137e in the SGB V.

1548 Huster, 'Der Gemeinsame Bundesausschuss als Ethikbehörde?' (2017) 35(4) MedR p. 282, 284. See also the position of the G-BA's chairman in an interview in Deckers and Mihm, '"Das wäre Zwei-Klassen-Medizin" Im Gespräch: Josef Hecken, Vorsitzender des Gemeinsamen Bundesausschusses' *Frankfurter Allgemeine Zeitung* (14.12.2016), p. 4.

1549 As pointed out by Huster, 'Der Gemeinsame Bundesausschuss als Ethikbehörde?' (2017) 35(4) MedR p. 282, 284.

1550 ibid. For details on the functioning of the reimbursement claim based on the so-called 'system failure', see Kingreen in Becker and Kingreen, *SGB V: Gesetzliche Krankenversicherung Kommentar* (7th edn 2020) para. 16-ff.

1551 Hufen, 'Zur verfassungsrechtlichen Beurteilung frühzeitiger pränataler Diagnostik', 4.1.2013, p. 17; Huster, 'Der Gemeinsame Bundesausschuss als Ethikbehörde?' (2017) 35(4) MedR p. 282, 285.

1552 Huster, 'Der Gemeinsame Bundesausschuss als Ethikbehörde?' (2017) 35(4) MedR p. 282, 284-ff.

1553 See ibid, p. 284.

1554 ibid.

Admittedly, there is indeed an abstract possibility for the legislature to provide a legal basis authorising or encouraging the use of ethical criteria in the G-BA decisions or in the health technology assessment conducted by the Institute for Quality and Efficiency in Health Care.[1555] Nonetheless, it must be considered that a series of constraints and precautions would be needed in order to guarantee that respect for constitutional standards is maintained.

In this respect, the G-BA's lack of democratic legitimacy[1556] already constitutes a disincentive to granting it the competence to decide which of the ethical and religious convictions, which are represented in the pluralist society, should prevail and contribute to the shaping of the publicly funded healthcare system.[1557] The inclusion of ethical concerns in health technology assessments would pave the way to taking into consideration extra-legal norms and standards that have neither undergone public discussion nor any democratic legitimation process. [1558] This would also result in the introduction of an element of arbitrariness into the decisions regarding the scope of the statutory health insurance.[1559]

Introducing ethicists amongst the members of the G-BA would not eliminate the problem either. While it is correct that ethicists can be considered experts in ethical argumentation and can show the flaws or strengths of certain arguments, it is also true that they cannot be legitimised to democratically represent the various moral and religious convictions that exist in a highly pluralistic society.[1560]

1555 As advocated, for instance, in Rüffer, 'Kostenübernahme für pränatale Bluttests. Pro und Contra' (2018) 114(44) Deutsches Ärzteblatt A1989, A1989.

1556 According to the Federal Constitutional Court, the doubts on the democratic legitimacy of the Federal Joint Committee shall be considered "quite weighty" (BVerfG, 10.11.2015 - 1 BvR 2056/12, author's translation), as highlighted by Kingreen, 'Der Gemeinsame Bundesausschuss vor dem BVerfG: Das Tor liegt in der Luft!' (2017) 35(1) MedR p. 8, 9.

1557 See Huster, 'Der Gemeinsame Bundesausschuss als Ethikbehörde?' (2017) 35(4) MedR p. 282, 285, who argues that a legal basis for the consideration of ethical concerns in the procedure would not be a viable option, given the already controversial legitimacy of the G-BA that would not benefit from such 'ethicalisation'.

1558 Gruschke in Vöneky and others, *Ethik und Recht - Die Ethisierung des Rechts/ Ethics and Law - The Ethicalization of Law* (2013) p. 42.

1559 ibid.

1560 Vöneky in Vöneky and others, *Legitimation ethischer Entscheidungen im Recht: Interdisziplinäre Untersuchungen* (2009).

But more fundamentally, the principle of the ethical neutrality of the state as a neutrality of justification comes into play. According to this standard, access to a health service could not be legitimately denied on the basis of purely ethical considerations.[1561]

As already mentioned, the legal assessment of NIPT does not differ compared to any other test for the prenatal diagnosis of fetal trisomies already available in the public healthcare system. From a social law perspective NIPT must be considered a valid innovation for the statutory health insurance. It meets the legal criteria set out in § 135(1) of the SGB V and fulfils the same function as invasive diagnosis, albeit without carrying any risk of miscarriage.

Also from the point of view of constitutional law NIPT raises no more concerns than existing invasive diagnoses. The balance between a woman's reproductive self-determination and the rights of the foetus does not change just because the diagnosis is less risky and therefore more widely used. [1562] In other words, the quantitative dimension of the use of prenatal diagnoses to detect fetal trisomies does not change their legal assessment.[1563]

Against this background, the only objection to the inclusion of NIPT in the statutory health insurance is an ethical one. Indeed, it can be argued that NIPT could contribute to increasing the use of prenatal screening. Indeed, with NIPT being reimbursed by health insurance funds, more women may potentially decide to take up the screening option, as this less invasive test presents no danger to the health of the foetus. A wider distribution of the diagnosis is considered by part of the society as ethically problematic, although it does not affect the legal and constitutional evaluation of the screening itself. Hence, any justification for refusing to reimburse NIPT through the statutory health insurance would only be grounded in ethical concerns related to the increased use of prenatal diagnoses. However, according to the principle of the ethical neutrality of the state, such concerns could not legitimately provide a basis of justification for measures taken

1561 See considerations on the ethical neutrality of the state in the German public healthcare system, in Chapter 1, sec. B.I.2.b.

1562 Heinrichs, Spranger and Tambornino, 'Ethische und rechtliche Aspekte der Pränataldiagnostik' (2012) 30(10) MedR p. 625, 629; Huber in Steger, Orzechowski and Schochow, *Pränatalmedizin* (2018) p. 155.

1563 Tolmein, 'Selbstbestimmungsrecht der Frau, Pränataldiagnostik und die UN-Behindertenrechtskonvention' (2012) 45(4) KJ p. 420, 430; Kießling in Rolfs and others, *BeckOK Sozialrecht* (2021) para. 11.

by the ethically neutral welfare state. In sum, the constitutional standard of neutrality of justification prevents the use of arguments drawn from specific ethical or religious convictions as legitimate criteria for the decisions of the G-BA.

B. Non-Invasive Prenatal Testing in Italy

I. NIPT in the Private Sector

Non-invasive prenatal testing entered the Italian private market through its CE marking in 2012.[1564]

Before being implemented in some Regional Healthcare Systems NIPT was only offered at several private clinics and laboratories with costs borne by the patients.[1565] Despite its initially high price, a study has found that uptake of NIPT in Italy was higher than the European average and its use occurred mainly through private clinics.[1566]

The rapid spread of NIPT in the private sector has caused some concerns. The Italian National Health Council (*Consiglio Superiore di Sanità*, CSS), the technical and scientific consulting body to the Ministry of Health, has been warning that some private facilities do not provide appropriate counselling before and after the test and has noted that patients have reported inadequate communication and informed consent.[1567] The CSS and other organisations have expressed their concern that the use of non-invasive screening for chromosomal trisomies predominantly in a deregulated private context would lead to biased reporting of scientific data and access to testing without the necessary quality assurance.[1568]

1564 At the time, entry into market of in vitro diagnostics with CE marking was regulated by d. lgs. n. 322/2000, as amended by d. lgs. 37/2010.

1565 Consiglio Superiore di Sanità, Sez. I, 'Linee-Guida. Screening prenatale non invasivo basato sul DNA (Non Invasive Prenatal Testing – NIPT)' (05.2015), p. 14 <https://www.salute.gov.it/imgs/C_17_pubblicazioni_2381_allegato.pdf> accessed 6.4.2022.

1566 Gadsbøll and others, 'Current Use of Noninvasive Prenatal Testing in Europe, Australia and the USA: A Graphical Presentation' (2020) 99(6) Acta Obstet Gynecol Scand p. 722, 724–725.

1567 Consiglio Superiore di Sanità, Sez. I, 'Linee-Guida. Screening prenatale non invasivo basato sul DNA (Non Invasive Prenatal Testing – NIPT)', 05.2015, p. 14.

1568 Fondazione ONDA, 'Atti tavolo tecnico interregionale Test Prenatali Non Invasivi (NIPT)' (Milano 13.12.2019), p. 5 <https://ondaosservatorio.it/ondauploads/20

Moreover, access to NIPT through the private market in the absence of homogeneous state funding has been noted to create inequalities both between different Regions across the national territory and between wealthy and less wealthy patients.[1569]

II. NIPT in the National Health Service

1. Access to Prenatal Screening and Diagnoses

a Prenatal Screening and Diagnoses in the Essential Levels of Care

In Italy screening and prenatal diagnosis procedures for chromosomal trisomies have long been part of the maternity protection measures contained in Essential Levels of Care.[1570] These represent the benefit basket of the National Health Service. As illustrated above,[1571] health services must be included in the LEA when they are necessary to guarantee the essential core of the fundamental right to health.[1572] As such they fall within the exclusive competence of the national legislature[1573] and must be equally provided to all national residents.[1574]

The inclusion of prenatal screening in the LEA thus indicates that they are considered part of a minimum standard of health protection that the state must ensure, as they are essential to the protection of the right to

20/10/NIPT-ONDA_atti-tavolo-tecnico_DEF.pdf> accessed 6.4.2022; Consiglio Superiore di Sanità, Sez. I, 'Linee-Guida. Screening prenatale non invasivo basato sul DNA (Non Invasive Prenatal Testing – NIPT)', 05.2015, p. 14.

1569 Fondazione ONDA, 'Atti tavolo tecnico interregionale Test Prenatali Non Invasivi (NIPT)', Milano 13.12.2019, p. 4; Consiglio Superiore di Sanità, Sez. I, 'Screening del DNA fetale non invasivo (NIPT) in sanità pubblica' (9.3.2021), p. 3 <https://www.salute.gov.it/imgs/C_17_pubblicazioni_3097_allegato.pdf> accessed 6.4.2022.

1570 See Decree of the Minister of Health of 10 September 1998 in Gazzetta Ufficiale no. 245 of 20.10.1998 also known as '*Decreto Bindi*'.

1571 See Chapter 1, sec. B.II.2.b.

1572 Pesaresi, 'La "determinazione dei livelli essenziali delle prestazioni" e la materia "tutela della salute": la proiezione indivisible di un concetto unitario di cittadinanza nell'era del decentramento instituzionale' (2006) 51(2) Giur Cost p. 1733, 1742.

1573 Art.117(2) letter m) Italian Constitution.

1574 Italian Constitutional Court, judgment no. 88/2003. See Balboni, 'I livelli essenziali e i procedimenti per la loro determinazione' [2003](6) Le Regioni p. 1183, 1187; Bergo, 'I nuovi Livelli Essenziali di Assistenza. Al crocevia fra la tutela della salute e l'equilibrio di bilancio' [2017](2) Rivista AIC p. 1, 5.

health of pregnant women. Prenatal diagnosis was already foreseen in the Decree of the Minister of Health of 10 September 1998 as an Essential Level of Care that was free of co-payment for certain categories of patients at risk.[1575]

In December 2015 an annex to the Decree of the Minister of Health containing the eligibility conditions for outpatient care services included an entry for the reimbursement of invasive tests, performed to confirm the finding of chromosomal trisomies detected by NIPT, by the National Health Service.[1576] However, NIPT itself was not covered, presumably because its clinical implementation was still at an early stage.

In 2017 the Prime Ministerial Decree of January 12th updated the catalogue of nationally provided health services. In its Article 59 the decree confirmed that prenatal diagnoses are nationally provided as part of the Essential Levels of Care.[1577] Moreover, the list of specialised outpatient services for pregnant women was updated by offering, for the first time, prenatal screening through combined testing free of charge to all patients.[1578] In the case of a high risk pregnancy, detected by the combined test or due to family conditions, invasive diagnoses would be offered regardless of the woman's age.[1579]

The DPCM of 12 January 2017 specified that the 2015 Decree of the Minister of Health on outpatient care services would stays in force until the approval of a new 'tariff decree', which at the time of writing has not yet been issued.[1580] Therefore, invasive tests confirming the result of NIPT remain nationally reimbursed.

1575 Decree of the Minister of Health of 10 September 1998 in Gazzetta Ufficiale no. 245 of 20.10.1998.

1576 Decree of the Minister of Health 9 December 2015 in Gazzetta Ufficiale no. 15 of 20.1.2016, attachment 2, p. 37. See Consiglio Superiore di Sanità, Sez. I, 'Screening del DNA fetale non invasivo (NIPT) in sanità pubblica', 9.3.2021, p. 4.

1577 DPCM 12 January 2017 in Gazzetta Ufficiale no. 65 of 18.3.2017, Suppl. n. 15. On the much awaited updating of the LEA, see, *inter alia,* Bergo, 'I nuovi Livelli Essenziali di Assistenza. Al crocevia fra la tutela della salute e l'equilibrio di bilancio' [2017](2) Rivista AIC p. 1; Vicarelli, 'I nuovi LEA: Passaggio storico o illusione collettiva?' [2017](3) Politiche Sociali p. 517.

1578 DPCM 12 January 2017 in Gazzetta Ufficiale no. 65 of 18.3.2017, Suppl. n. 15, attachment 10B.

1579 ibid attachment 10C.

1580 This decree is still applicable pending the decree defining the maximum tariffs for ambulatory services, see Art. 64(2) DPCM 12 January 2017 in Gazzetta Ufficiale no. 65 of 18.3.2017, Suppl. n. 15. Concrete steps towards the adoption of this decree were only taken at the end of January 2022, see Martini and Marchetti, 'Decreto

However, there is still no mention of NIPT in the updated regulation containing the new Essential Levels of Care. Yet this omission does not seem to stem from an ideological opposition against NIPT. The Italian public debate on NIPT was in fact nowhere near as extensive as in Germany and the UK. As will be illustrated in detail below,[1581] government and parliamentary bodies have rather unanimously insisted on the benefits of non-invasive diagnoses.

From the perspective of legal scholars, the few contributions published on this question have primarily called for a careful consideration of informed consent issues in the possible implementation of NIPT in clinical practice.[1582] It is considered that the increased availability of these non-invasive testing methods will add significantly to the patients' need for accurate and unbiased information.[1583] It is considered essential, *inter alia*, that women have a realistic option of deciding not to undergo any kind of prenatal screening and that they are made aware of alternatives to abortion.[1584] Particular concerns are only voiced in view of the possible use of NIPT to detect non-pathological features in the foetus, such as aesthetic traits or other non-medical conditions.[1585]

Doubts were also expressed about the possible routinisation of NIPT use and a perceived stigmatisation of the community of people with disability

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sulle tariffe e aggiornamento dei LEA: una neverending story?' *Quotidiano Sanità* (8.2.2022) <https://www.quotidianosanita.it/lettere-al-direttore/articolo.php?articolo_id=102142> accessed 6.4.2022. This problematic delay prevents the new services from being offered at the expense of the National Health System, and creates inequalities especially against Regions subject to recovery plans, which cannot implement the new LEAs on their own.

1581 See below in this section at para. II.3.

1582 Palazzani, *Dalla bio-etica alla tecno-etica: Nuove sfide al diritto* (2017) pp. 138–144; Rizzo, 'Il consenso informato come strumento per l'implementazione etica dei test genetici non invasivi per la diagnosi prenatale' [2018](3) BioLaw Journal – Rivista di BioDiritto p. 225.

1583 Palazzani, *Dalla bio-etica alla tecno-etica* (2017) p. 142; Rizzo, 'Il consenso informato come strumento per l'implementazione etica dei test genetici non invasivi per la diagnosi prenatale' [2018](3) BioLaw Journal – Rivista di BioDiritto p. 225, 229.

1584 Palazzani, *Dalla bio-etica alla tecno-etica* (2017) p. 144; Rizzo, 'Il consenso informato come strumento per l'implementazione etica dei test genetici non invasivi per la diagnosi prenatale' [2018](3) BioLaw Journal – Rivista di BioDiritto p. 225, 229.

1585 Rizzo, 'Il consenso informato come strumento per l'implementazione etica dei test genetici non invasivi per la diagnosi prenatale' [2018](3) BioLaw Journal – Rivista di BioDiritto p. 225, 232–234.

and women deciding to avoid testing.[1586] In this regard, the question of whether NIPT should be provided to patients through public funding was raised, although it was concluded that adequate information and communication with patients before and after the test would overcome possible doubts.[1587]

In short, it seems that most of the contributors to the Italian debate maintain that the new moral issues emerging with NIPT can be resolved by means of adequate counselling, provided that this is realised in practice. Except for calls for the consideration of possible informed consent issues,[1588] there has not been much debate in the wider public sphere about the desirability of NIPT in general. NIPT seems to be tacitly accepted as an improvement in the safety and accuracy of previous diagnostic techniques.

b Informed Consent

The understanding of prenatal diagnoses as part of a minimum standard of health that the state must protect is in line with the principles endorsed by the Italian Constitution. This places a very high value on the right to health and the right to self-determination in matters of health, according to the combination of Articles 2, 13 and 32. In particular, information on the health condition of the foetus is considered, both by legislation and the case law, to be closely connected with the physical and psychological health of pregnant women.[1589] The Court of Cassation pointed out that prenatal diagnosis is relevant to a woman's health not only insofar as it enables her to make an abortion decision but also because, if abortion is ruled out, it

1586 Rizzo, 'Il consenso informato come strumento per l'implementazione etica dei test genetici non invasivi per la diagnosi prenatale' [2018](3) BioLaw Journal – Rivista di BioDiritto p. 225, 234.

1587 Rizzo, 'Il consenso informato come strumento per l'implementazione etica dei test genetici non invasivi per la diagnosi prenatale' [2018](3) BioLaw Journal – Rivista di BioDiritto p. 225, 239.

1588 Fondazione ONDA, 'Atti tavolo tecnico interregionale Test Prenatali Non Invasivi (NIPT)', Milano 13.12.2019, p. 14.

1589 Article 6 Law no. 194/1978 and Corte di Cassazione, judgments nos. 16754/2012, 25767/2015 and 5004/2017. See Conte, '"And makes us rather bear those ills we have?" L'inizio della vita e i confini della sofferenza risarcibile (Nota a Corte di Cassazione, Sezioni Unite, n. 25767/2015)' [2016](2) BioLaw Journal – Rivista di BioDiritto p. 433, 436.

equips her psychologically and materially for the birth of a child with a particular genetic condition.[1590]

The right to self-determination and the right to health are brought together under the umbrella principle of informed consent, according to which each patient has the right to receive information on their health status and on the available medical possibilities in order to be able to make a free and informed choice.[1591] The right to informed consent received dedicated statutory protection with Law no. 219/2017, which placed a special focus on doctor-patient dialogue by establishing that the time spent in communication effectively constitutes treatment time.[1592]

In the case of prenatal screening, the right to informed consent must be read in conjunction with the constitutional requirement of laicity of the State.[1593] In adopting a laicity-driven approach the ethical perception of the woman and the foetus occupying essentially conflicting positions must be abandoned and the woman's right to prenatal diagnoses must be seen as the result of a balancing of rights in compliance with the relevant constitutional principles.[1594]

1590 Corte di Cassazione, judgment no. 5004/2017. See Salvatore, 'La recente legge sul consenso informato. Un passo in avanti in tema di responsabilità medica per violazione degli obblighi informativi?' [2018](3) Riv ital med leg dirit campo sanit p. 993, 1007.

1591 The concept of informed consent as grounded in Articles 2, 13 and 32 of the Constitution was elaborated for the first time in the Italian Constitutional Court judgment no. 438/2008. This was recently confirmed in the Italian Constitutional Court judgment no. 144/2019, see Balduzzi and Paris, 'Corte costituzionale e consenso informato tra diritti fondamentali e ripartizione delle competenze legislative' (2008) 53(6) Giur Cost p. 4953; Casonato, 'Il principio della volontarietà dei trattamenti sanitari fra livello statale e livello regionale: Nota a Sentenza n. 438/2008' (2009) 37(3-4) Le Regioni p. 627, 627–628.

1592 Russa and others, 'Consenso informato e dat (disposizioni anticipate di trattamento): Momento legislativo innovativo nella storia del biodiritto in italia' (2018) 83(1) Responsabilità civile e previdenza p. 353, 359; Salvatore, 'La recente legge sul consenso informato. Un passo in avanti in tema di responsabilità medica per violazione degli obblighi informativi?' [2018](3) Riv ital med leg dirit campo sanit p. 993, 996–997.

1593 D'Amico, 'Il concepito e il diritto a nascere sani: Profili costituzionali alla luce della decisione della Corte di Cassazione (n. 16754 del 2012)' [2014](2) Rivista AIC p. 1, 2.

1594 As envisaged in the abortion decision of the Italian Constitutional Court, judgment no. 27/1975; see D'Amico, 'Il concepito e il diritto a nascere sani: Profili costituzionali alla luce della decisione della Corte di Cassazione (n. 16754 del 2012)' [2014](2) Rivista AIC p. 1, 5; Conte, '"And makes us rather bear those ills we have?" L'inizio della vita e i confini della sofferenza risarcibile (Nota a Corte

In the light of these principles, prenatal screening for chromosomal trisomies has traditionally been seen as relatively uncontroversial in Italy, provided that the patient's fully informed consent is maintained. As early as 1992 the Italian Committee for Bioethics (*Comitato Nazionale per la Bioetica*, CNB) issued an "overall positive"[1595] assessment of the different prenatal screening procedures and argued that the right to know the health status of the foetus was undisputed.[1596] However, the document stressed that couples should be provided with a 'non-directive' medical consultation, i.e. the information given by the doctor should not exert any pressure to undergo the diagnosis and the doctor should refrain from encouraging or discouraging abortion.[1597] The members of the Committee noted that the permissibility of prenatal diagnoses could be challenged only if they were associated with selection and eugenic purposes.[1598] In this regard, it was emphasised that prenatal screening should be kept conceptually distinct from any possible abortion choice. The CNB recommended that the essential distinction between the two moments must be guaranteed in practice and borne in mind during the consultation.[1599]

2. Coverage of NIPT in Different Regional Healthcare Systems

As it is not currently included in the Essential Level of Care, public funding of NIPT is still left to the discretion of individual Regional Healthcare Systems.

Regions have, first of all, the task of implementing the Essential Level of Care in their Regional Healthcare Systems. As regards prenatal screening, annex 10C of the Prime Minister's Decree of 12 January 2017 calls on the Regions to adopt methods for calculating the risk of chromosomal trisomies in pregnancy that have greater sensitivity and fewer false positives, taking into account the developments in scientific research.

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di Cassazione, Sezioni Unite, n. 25767/2015)' [2016](2) BioLaw Journal – Rivista di BioDiritto p. 433, 436.

1595 Comitato Nazionale per la Bioetica, 'Diagnosi prenatali', 18.7.1992, p. 28 (author's translation).

1596 ibid, pp. 36-37.

1597 ibid, pp. 30-31.

1598 ibid, p. 42.

1599 ibid, p. 43.

Moreover, individual Regions have the possibility to include additional so-called 'extra-LEA' services in their Regional Healthcare System's services.[1600] While the essential levels of protection must be guaranteed throughout the national territory,[1601] the Regions have concurrent legislative competence in the sphere of health protection according to Article 117(3) of the Italian Constitution.[1602] This increase in benefits may be offered in line with the political orientation of each Region and by allocating funds from the regional budget.[1603]

On the basis of their concurrent competence, and in light of the persistent delay of the national government, many Regions have decided to independently undertake action to publicly fund and provide NIPT to their residents. This development was also prompted by the 2015 guidelines of the Italian National Health Council, which recommended the introduction of NIPT in all public facilities.[1604]

Emilia Romagna has decided to offer free NIPT to all pregnant women regardless of risk factors. Already in March 2015 this Region commissioned a scientific evaluation on the possibility of including NIPT in the Region's antenatal pathway.[1605] The assessment team also included members with

1600 See Pellegrini in Balduzzi, *La sanità italiana tra livelli essenziali di assistenza, tutela della salute e progetto di devolution: Atti del convegno, Genova, 24 febbraio 2003* (2004). The possibility for Regions to offer additional health services to their residents is an entirely physiological feature of the Italian public healthcare system. However, the concentration of therapeutic and diagnostic innovations in only a few Regions leaves room for potentially unsustainable inequalities, see Aperio Bella, 'Tecnologie innovative nel settore salute tra scarsità delle risorse e differenziazione: alla ricerca di un equilibrio difficile' [2020](2) Federalismi p. 245, 260–261.

1601 Art. 117(2) letter m) Italian Constitution.

1602 Regions have legislative powers in all matters of concurrent legislation, except for the determination of fundamental principles, which is reserved for State legislation, see Art. 117(3) Italian Constitution.

1603 Balboni, 'I livelli essenziali e i procedimenti per la loro determinazione' [2003](6) Le Regioni p. 1183, 1191.

1604 Consiglio Superiore di Sanità, Sez. I, 'Linee-Guida. Screening prenatale non invasivo basato sul DNA (Non Invasive Prenatal Testing – NIPT)', 05.2015. See, for instance, references to the CSS guidelines in Regione Emilia-Romagna (Giunta Regionale), Delibera no. 1894, 4.11.2019; Regione Umbria (Assemblea Legislativa), Deliberazione no. 279, 23.10.2018.

1605 Gruppo di Lavoro Regionale Test Prenatali Non Invasivi (NIPT), 'Resoconto delle attività: marzo - giugno 2015' (28.12.2015), p. 5 <https://assr.regione.emilia-romagna.it/pubblicazioni/rapporti-documenti/test-prenatali-2015/@@download/publicationFile/Gruppo%20RER%20NIPT.pdf> accessed 6.4.2022; Fondazione

ethical and legal expertise and citizen representatives and thus assessed not only the clinical and organisational implications but also the ethical and legal consequences of offering NIPT in the Regional Healthcare System.[1606] This group found a unanimous consensus in recommending the offer of NIPT as a replacement for the combined test.[1607] Accordingly, in 2019 the Region's governing body gave its approval to the Regional Healthcare System offering NIPT to all pregnant women for the detection of the presence of trisomy 12, 18 and 21.[1608] This was initially launched as a 9-month trial project at the end of which an evaluation and subsequent confirmation of the new screening pathway would be carried out.[1609]

In the self-governing province of Bolzano the provincial government decided to offer NIPT at the expense of the provincial health service at the end of 2018, albeit only to patients who, following the assessment of the combined test, were found to be at intermediate risk of having an affected foetus.[1610] The provincial deliberation stipulated that women who meet the requirements to be eligible for the test should be given an informational consultation aimed at guiding the patient to an informed choice and at collecting informed consent. This counselling is granted an independent tariff and reimbursement code.[1611]

In late 2018 Tuscany also decided to integrate NIPT into the catalogue of specialised outpatient services provided by the Regional Healthcare System.[1612] Here this test is available to pregnant women who have been found to be at risk of between 1/301 and 1/1000 after the combined test. [1613] Unlike Emilia Romagna and Bolzano, however, Tuscany asks for a patient

ONDA, 'Atti tavolo tecnico interregionale Test Prenatali Non Invasivi (NIPT)', Milano 13.12.2019, p. 10.

1606 Gruppo di Lavoro Regionale Test Prenatali Non Invasivi (NIPT), 'Resoconto delle attività: marzo - giugno 2015', 28.12.2015, p. 29.

1607 Fondazione ONDA, 'Atti tavolo tecnico interregionale Test Prenatali Non Invasivi (NIPT)', Milano 13.12.2019, p. 10.

1608 Regione Emilia-Romagna (Giunta Regionale), Delibera no. 1894, 4.11.2019, Art. 1.

1609 ibid Art. 2.

1610 Provincia Autonoma di Bolzano - Alto Adige (Giunta Provinciale), Deliberazione no. 1413, 18.12.2018, p. 31.

1611 ibid p. 32.

1612 Regione Toscana (Giunta Regionale), Delibera no. 1371, 10.12.2018. See also 'Percorso nascita. In Toscana test combinato gratuito a tutte le gestanti e test Nipt a tariffa ridotta. Saccardi: "Facciamo da apripista a livello nazionale"' *Quotidiano Sanità* (5.3.2019) <https://www.quotidianosanita.it/regioni-e-asl/articolo.php?articolo_id=71605> accessed 6.4.2022.

1613 Regione Toscana (Giunta Regionale), Delibera no. 1371, 10.12.2018.

co-payment amounting to half the price. Full reimbursement of costs is only granted to pregnant women who fall into certain high risk or low income categories.[1614]

In 2021, the parliamentary body of the Region Puglia unanimously approved a bill to provide NIPT as prenatal screening for the detection of chromosomal trisomies to pregnant women over the age of forty or those who are found to be at a high or intermediate risk after combined testing.[1615] The main aim of the bill was to improve the quality of pregnancy in both medical and psychological terms and to limit the risks of invasive diagnosis.[1616]

The enactment of this regional law was challenged by the Italian government before the Constitutional Court.[1617] The central government argued that the introduction of NIPT into the Regional Healthcare System in Puglia is in breach of the financial deficit recovery plan to which the Region is subject.[1618] The appeal before the Constitutional Court on the regional law on public funding of NIPT is currently pending.

1614 ibid.

1615 Art. 3 Legge Regionale Puglia no. 31/2021, "Implementazione del Test prenatale non invasivo (NIPT)" 6.8.2021

1616 Articles 1 and 3 Legge Regionale Puglia no. 31/2021, "Implementazione del Test prenatale non invasivo (NIPT)" 6.8.2021.

1617 Ricorso per legittimità costituzionale 6.10.2021, in Gazzetta Ufficiale 1° Serie Speciale (Corte Costituzionale) no. 43 of 27.10.2021, N. 55, p. 62 < https://www.gazzettaufficiale.it/atto/corte_costituzionale/caricaDettaglioAtto/originario?atto.dataPubblicazioneGazzetta=2021-10-27&atto.codiceRedazionale=21C00246> accessed 10.8.2022.

1618 Ricorso per legittimità costituzionale 6.10.2021, in Gazzetta Ufficiale 1° Serie Speciale (Corte Costituzionale) no. 43 of 27.10.2021, N. 55, p 62. Recovery plans are an instrument through which the national legislature ensures that Regions in a financial deficit maintain the provision of the Essential Levels of Care and contain public health expenditure. The introduction of a recovery plan is admittedly a fairly significant level of State interference in the sphere of regional autonomy. However, it is justified by the exclusive competence of the State in relation to the coordination of public finance and the determination of the essential levels of services that must be guaranteed to all residents across the national territory, see Carpani in Balduzzi, *La sanità italiana alla prova del federalismo fiscale* (2012) pp. 36-37; Cerioni, 'Stato e Regioni di fronte alla gestione dei Piani di rientro nei sistemi sanitari regionali in deficit' [2017](1) Politiche Sociali p. 175, 176. As confirmed also recently by a ruling of the Constitutional Court against Puglia, the Regions subject to the recovery plans cannot foresee additional expenses to those necessary to guarantee the LEA, see Italian Constitutional Court, judgment no. 142/2021, considerations in point of law para. 2.

In Basilicata too, a project was approved in 2019 to include NIPT in the prenatal screening pathway in order to reduce the number of invasive diagnoses performed. Also in this Region the test will be offered to pregnant women at intermediate risk after the combined test.[1619]

In November 2021, Lombardia started a 6-month phase of provisional reimbursement in which NIPT is offered in one regional health facility before being opened up for all pregnant women in the Region.[1620]

A few more Regions, although they have not yet included NIPT in their regional health benefit catalogues, have at least initiated its assessment or expressed political will in this direction.

In Umbria this occurred as early as 2018 when the legislative assembly unanimously passed a resolution committing the regional governing body to consider introducing NIPT for chromosomal trisomies for all women over the age of 35.[1621] The main aim was stated to be the reduction of potential, albeit rare, harm resulting from the use of invasive diagnosis among women over 35.

The regional legislative assembly of Piemonte followed in 2021, when it issued an agenda committing the regional government to consider the introduction of NIPT for all women regardless of their age and risk factors.[1622] This political motion was approved almost unanimously by the governmental majority and the opposition[1623] and was at least on one occasion criticised in the local press out of concern that the inclusion of non-invasive tests for chromosomal trisomies in the Regional Healthcare System would create a slippery slope towards stigmatisation of people with disabilities.[1624]

Also in 2021, Liguria's parliamentary body unanimously issued an agenda calling on the regional government to consider including NIPT free of

1619 Regione Basilicata (Giunta Regionale), Delibera no. 456, 12.7.2019.

1620 'Lombardia. Approvati nuovi test fetali non invasivi per le donne in gravidanza' *Quotidiano Sanità* (16.11.2021) <http://www.quotidianosanita.it/regioni-e-asl/articolo.php?articolo_id=100054> accessed 6.4.2022.

1621 Regione Umbria (Assemblea Legislativa), Deliberazione no. 279, 23.10.2018.

1622 Regione Piemonte (Consiglio Regionale), Ordine del giorno no. 170, 3.2.2021.

1623 Giacosa, 'Sinistra e Lega, la "strana coppia" che in Piemonte ha ottenuto il test del dna per le donne incinte' *La Repubblica* (4.2.2021) <https://torino.repubblica.it/cronaca/2021/02/04/news/strana_coppia_sinistra_lega_test_dna_gravidanza_gratuito-285999290/> accessed 6.4.2022.

1624 Dovico, 'Il Piemonte, il Nipt e la china scivolosa sui bimbi Down' *La Nuova Bussola Quotidiana* <https://lanuovabq.it/it/il-piemonte-il-nipt-e-la-china-scivolosa-sui-bimbi-down> accessed 6.4.2022.

charge in the regional antenatal diagnosis pathway.[1625] The government accepted this invitation and set up a technical panel in August 2021 to evaluate the test.[1626]

3. Prospective Coverage of NIPT at the National Level

a Guidelines of the Italian National Health Council

While several Regions have already taken action, parliamentary and governmental bodies at the national level have correctly pointed out that NIPT must be implemented by the National Health Service, claiming that it belongs to the minimum services that the state must provide to all residents in order to protect the essential core of the right to health.

In May 2015, the Italian National Health Council issued its first guidelines on NIPT. The document was drafted by the first section of the Council, which is responsible, *inter alia*, for consulting the Ministry of Health on the Essential Levels of Care and on HTA for the evaluation of innovative technologies in the National Health Service.[1627] The multidisciplinary team working on the NIPT recommendations included three members of the CNB.[1628]

The guidelines maintained that NIPT would not provide an incentive for inappropriate use of prenatal screening compared to current clinical practice. If only used for trisomies 21, 18 and 13 it would not expand the range of conditions for which many women already wish to be informed.[1629] In this respect, one benefit of NIPT would be that it provides more accurate

1625 'Nipt test. Regione valuta utilizzo gratuito dopo Odg approvato all'unanimità in Consiglio' *Quotidiano Sanità* (7.4.2021) <https://www.quotidianosanita.it/liguria/articolo.php?articolo_id=94373> accessed 6.4.2022.

1626 Azienda Ligure Sanitaria della Regione Liguria, Deliberazione no. 308, 11.8.2021.

1627 Art. 7 Decree of the Minister of Health, 6 August 3002, n. 342 Gazzetta Ufficiale no. 287 of 11.12.2003. The functions of the CSS are laid down in Article 4 of d.lgs. no. 266/1993, according to which the *Consiglio Superiore di Sanità* may, among other things, propose the study of problems relating to hygiene and health and propose to the health administration the formulation of draft rules and measures for the protection of public health.

1628 The list of the members of the working group is available in Consiglio Superiore di Sanità, Sez. I, 'Linee-Guida. Screening prenatale non invasivo basato sul DNA (Non Invasive Prenatal Testing – NIPT)', 05.2015, p. 3.

1629 ibid, p. 13.

information without putting the pregnancy at risk.[1630] To this end, however, the Council argued that it is essential to make counselling an integral part of the screening offer and to thoroughly prepare prospective parents for the information that will result from the test and the possible decisions to be taken as a consequence.[1631] In addition, the guidelines acknowledged that new ethical issues may arise if the number of detectable genetic conditions would increase.[1632]

In conclusion, the CSS supported the need for NIPT to be offered by the healthcare systems at central and regional level.[1633] Centralisation of testing laboratories was also recommended to maintain cost-effectiveness.[1634]

In a follow-up paper in 2016 another working group of the National Health Council assessed the socio-economic impact of incorporating NIPT into public healthcare. This argued that establishing criteria and modalities for testing at a national level would be necessary to overcome some critical issues related to its unregulated use in the private sector.[1635] The sensitive nature of the issue was pointed out in relation to the ethical, emotional and social implications of NIPT and its connection with abortion. [1636] The working group concluded by recommending that the National Health Service should reimburse NIPT as a contingent addition to the combined test, whereby the service must be subject to adequate standards of quality and proper informed consent mechanisms.[1637]

Building on the Council's guidelines, an agreement between the Regions and the State in October 2017 recommended promoting country-wide implementation of NIPT as one of the priorities for the innovation of the National Health Service.[1638]

1630 ibid.
1631 ibid.
1632 ibid.
1633 ibid, p. 19.
1634 ibid, p. 15.
1635 Consiglio Superiore di Sanità, Sez. I, 'Gruppo di Lavoro "NIPT 2". Impatto socio-economico del test del cfDNA/NIPT in Sanità pubblica' (07.2016), p. 12 <http://www.plurigentest.it/NIPT2%20%20doc%20%20finale%2012%20LUGLIO%202016.pdf> accessed 6.4.2022.
1636 ibid, p. 13.
1637 ibid, passim.
1638 Conferenza Stato-Regioni, 'Intesa, ai sensi dell'articolo 8, comma 6, della legge 5 giugno 2003, n. 131, tra il Governo, le Regioni e le Province autonome di Trento e Bolzano sul documento recante "Piano per l'innovazione del sistema sanitario basata sulle scienze omiche"' (26.10.2017).

The CSS last confirmed and updated its recommendations regarding NIPT in March 2021. In this document the Council noted with concern the inequalities created by devolving the implementation of NIPT to individual Regions.[1639] The main purpose of the new guidelines was therefore to recommend national implementation of NIPT. It was suggested that NIPT should be included in the Essential Levels of Care as contingent screening for trisomies 13, 18 and 21 after combined testing.[1640]

More space was devoted in these revised guidelines to the ethical considerations involved in the implementation of the test. For instance, the conflict between liberalist theories, utilitarian approaches and dignitarian perspectives is mentioned.[1641] The ethical problems of prenatal screening appear to be accentuated by the availability of extensive information about the foetus through a simple blood sample.[1642] The ease with which the test can be carried out could lead to the risk that pregnant women undergo screening without previous adequate critical reflection.[1643] To overcome such ethical concerns, the Council once again emphasised the importance of counselling and the provision of information on alternatives and consequences of the test as an integral part of screening. It reiterated that counselling should be 'non-directive' and that the couple should be able to freely choose whether to undergo screening or not.[1644]

The document also indicated as ethically problematic the possibility that the widespread use of the test would lead to increased discrimination or social exclusion against people with disabilities.[1645] As a response to this issue it is argued that the inclusion of NIPT in the offer of the National Health Service must be accompanied by policies of social justice and support for people with disabilities.[1646]

1639 Consiglio Superiore di Sanità, Sez. I, 'Screening del DNA fetale non invasivo (NIPT) in sanità pubblica', 9.3.2021, p. 3.
1640 ibid, p. 5.
1641 ibid, p. 19.
1642 ibid, p. 20.
1643 ibid.
1644 ibid, p. 21.
1645 ibid.
1646 ibid, p. 22.

b NIPT before the National Commission for the Updating of the Essential Levels of Care

At the end of 2019, on the occasion of the budget bill for the financial year 2020, the Health Commission of the Senate presented an order of the day (*ordine del giorno*) committing the government[1647] to "assess the possibility of taking initiatives to include NIPT in the Essential Levels of Care".[1648] The Commission considered it necessary to ensure equal access throughout the country to safer and more accurate prenatal screening procedures.[1649] The government accepted this order of the day, thereby making a political commitment to explore the possibility of national public funding for NIPT.

In the same year the Ministry of Health received a parliamentary question from a member of the Senate urging it to undertake initiatives to include NIPT in the Essential Levels of Care, also in the light of the guidelines of the National Health Council.[1650]

In response to these political solicitations the Ministry for Health confirmed that the proposal to include NIPT in the outpatient specialised services of the National Health Service would be submitted to the National Commission for the Updating of Essential Levels of Care.[1651] The Ministry announced that the decision on the inclusion of NIPT in the LEA would

1647 In Italian parliamentary law, an order of the day is an instrument with which Parliament exercises its political steering function vis-à-vis the government. The submission of orders of the day in connection with the draft budget law is governed by a special procedure in the parliamentary regulations for the Senate see Art. 127(1). An order of the day that 'commits' the government to a certain action is more binding than one that merely 'invites' it, although it is still only politically and by no means legally binding. The degree of the binding political force of the order of the day increases if the government fully accepts it. On the orders of the day in Italian parliamentary law, see Ciaurro, 'Ordine del giorno' (1980) XXX Enc dir p. 1018, 1035–1037; Mannino and Curreri, *Diritto parlamentare* (2019) p. 316-ff.

1648 Senato della Repubblica, 'Ordine del Giorno n. G/1586 sez I/18/12 (testo 2) al DDL n. 1586' <https://www.senato.it/japp/bgt/showdoc/frame.jsp?tipodoc=Emendc&leg=18&id=1126780&idoggetto=1134832> accessed 6.4.2022, author's translation.

1649 ibid.

1650 Senato della Repubblica, 'M. Rizzotti. Legislatura 18 Atto di Sindacato Ispettivo n° 3-01021' <https://www.senato.it/japp/bgt/showdoc/18/Sindisp/0/1118781/index.html> accessed 6.4.2022.

1651 'Test prenatali. Sileri: "All'esame della Commissione Lea inserimento dei test non invasivi"' *Quotidiano Sanità* (9.1.2020).

be taken in cooperation with the Regions and with the involvement of technical and scientific bodies, including the CSS.[1652]

c Criteria for Updating the Essential Levels of Care and Room for Ethical Concerns

A first Commission with the task of updating the Essential Levels of Care was already founded by Decree-Law no. 63 of 15 April 2002.[1653] Later on this function was taken over by the Technical Committee for Health operating at the Ministry of Health.[1654] Currently the updating of the LEA catalogue is entrusted to the National Commission for the Updating of Essential Levels of Care, established by Article 1(556) of Law 208/2015. The Commission has the task of systematically evaluating the health care services included in the LEA in order to decide on the maintenance of already existing measures and on proposals for the inclusion of new benefits in the catalogue.[1655] It is envisaged that on this basis the commission should formulate an annual proposal for the updating of the Essential Levels of Care, which would then be implemented by a decree either of the Minister of Health, if no additional costs arise for the public budget, or of the President of the Council of Ministers.[1656]

The legal framework governing the updating of the LEA lays down precise rules on the criteria to be used when assessing new health technologies. As provided for in Article 1(2) of Legislative Decree no. 502 of 30 December 1992, the Essential Levels of Care must be designed in accordance with the principles of human dignity, healthcare needs, equal access, quality of care, appropriateness and economical use of resources.[1657] In order for a new health service to be included in the benefit basket it must comply with

1652 ibid.

1653 Article 4-bis(10) Decree-law 63/2002, see Bergo, 'I nuovi Livelli Essenziali di Assistenza. Al crocevia fra la tutela della salute e l'equilibrio di bilancio' [2017](2) Rivista AIC p. 1, 8–9.

1654 Decree of the President of the Republic, 28 March 2013, n. 44 Gazzetta Ufficiale no. 98 of 27.4.2013.

1655 Art. 1(557) Law no. 208/2015.

1656 Respectively Art. 1(559) and Art. 1(554) of Law no. 208/2015. On how the commission operates in general see Bergo, 'I nuovi Livelli Essenziali di Assistenza. Al crocevia fra la tutela della salute e l'equilibrio di bilancio' [2017](2) Rivista AIC p. 1, 8-9; Vicarelli, 'I nuovi LEA' [2017](3) Politiche Sociali p. 517, 519.

1657 As amended by Article 1 d. lgs. no. 229/1999.

these guiding principles and meet requirements of appropriateness.[1658] The statutory text affirms that, in order to meet the appropriateness criterion, the effectiveness of the health service must be proven on the basis of scientific evidence.[1659]

The appropriateness criterion is a cornerstone principle in this framework.[1660] On the one hand, clinical appropriateness implies quality and safety of health services[1661] and in particular that the benefits for the patient should outweigh the risks.[1662] On the other hand, appropriateness also means that the catalogue of services must be constantly updated, taking into account innovative scientific developments.[1663] The wording of the legislation states that the purpose of the Commission is to ensure the effectiveness and clinical appropriateness of the services provided by the National Health Service, also in relation to scientific and technological developments.[1664] In other words, the appropriateness of a health technology is measured by its compliance with constantly developing scientific and technical rules in response to which the Essential Level of Care must also be adjusted.[1665]

Given the principles guiding the definition and updating of the Essential Levels of Care it would not seem that the Commission is entitled to consider possible ethical issues regarding individual technologies in the updating

1658 Art. 1(7) d. lgs. no. 502/1992, as amended by Article 1 d.lgs. no. 229/1999.

1659 Art. 1(7) letter b) d.lgs. no. 502/1992, as amended by Article 1 d.lgs. no. 229/1999; see Antonelli, 'La garanzia dei livelli essenziali di assistenza nei primi 40 anni del Servizio sanitario nazionale: dall'uniformità all'appropriatezza: efficacia non è dimostrabile in base alle evidenze scientifiche' [2018](7) Federalismi p. 1, 19.

1660 Molaschi, 'Sulla determinazione dei livelli essenziali delle prestazioni: riflessioni sulla vis expansiva di una 'materia'' [2003](5) Sanità Pubblica e Privata p. 525, 538; Antonelli, 'La garanzia dei livelli essenziali di assistenza nei primi 40 anni del Servizio sanitario nazionale: dall'uniformità all'appropriatezza: efficacia non è dimostrabile in base alle evidenze scientifiche' [2018](7) Federalismi p. 1, 19.

1661 Antonelli, 'La garanzia dei livelli essenziali di assistenza nei primi 40 anni del Servizio sanitario nazionale: dall'uniformità all'appropriatezza: efficacia non è dimostrabile in base alle evidenze scientifiche' [2018](7) Federalismi p. 1, 21–22.

1662 Materia, 'Appropriatezza: Origini, implicazioni, valutazione' [2003](4-5) Tendenze nuove p. 343, 344.

1663 Pesaresi, 'La "determinazione dei livelli essenziali delle prestazioni" e la materia " tutela della salute": la proiezione indivisible di un concetto unitario di cittadinanza nell'era del decentramento instituzionale' (2006) 51(2) Giur Cost p. 1733, 1760.

1664 Art. 1(556) Law no. 208/2015.

1665 Pesaresi, 'La "determinazione dei livelli essenziali delle prestazioni" e la materia " tutela della salute": la proiezione indivisible di un concetto unitario di cittadinanza nell'era del decentramento instituzionale' (2006) 51(2) Giur Cost p. 1733, 1757.

process.[1666] The Commission should not refuse to include an innovation in the benefit basket if this was, first, proven to be necessary to protect the minimum core of the right to health and, second, in line with the criterion of appropriateness.[1667] The constitutional framework governing the LEA is intended precisely to protect the essential content of the right to health against political determinations or ethical and religious influences on the legislature or government.[1668]

Nevertheless, the former Commission for the updating of the LEA, established in 2002, had explicitly included the consideration of ethical aspects in its methodology for the assessment of new health technologies. The strategy designed by the Commission consisted of a series of questions aimed at ascertaining whether a new health technology could be included in the Essential Levels of Care.[1669] The questions were largely drafted on the basis of the normative criteria of appropriateness and efficient use of resources, but also took into account further aspects. Among these additional factors, one of the questions that the Commission identified for its assessment concerned the 'ethical desirability' of the service. The question read: "Is this a service that is manifestly at odds with the fundamental ethical principles of our society?".[1670]

1666 This is also reflected in the purely 'technical' composition of this commission, which is chaired by the Minister of Health and composed of the Director of the Directorate-General for Health Planning of the Ministry of Health, fifteen qualified experts and the same number of substitutes, four of whom are designated by the Minister of Health, one by the Italian National Institute of Health, one by the Agenas, one by Aifa, one by the Ministry of the Economy and Finance and seven by the Conference of Regions and Autonomous Provinces, see Article 1(556) Law no. 208/2015.

1667 NIPT appears to comply with this description fully. First of all, as a method of prenatal diagnosis, it falls within the essential scope of the right to health that the state must guarantee. Secondly, NIPT improves the accuracy, safety and quality of prenatal diagnosis, thus constituting an innovation fulfilling the standard of appropriateness.

1668 For the illustration of the conception of the Essential Levels of Care as a guarantee of the minimum core of the right to health against possible political determinations of the state, see Chapter 1, sec. B.II.2.b.

1669 Commissione nazionale per la definizione e l'aggiornamento dei Livelli essenziali di assistenza in Falcitelli and Langiano, *La remunerazione delle attività sanitarie: Caratteristiche attuali e ipotesi evolutive* (2007) pp. 232-ff; Arcà and Cislaghi, 'Percorsi metodologici per l'inserimento o l'esclusione di una prestazione dai Livelli essenziali di assistenza' [2006](2) Tendenze nuove p. 97, 98-ff.

1670 (Autor's translation). Ordered according to their power of exclusion, the question of the ethical nature of the service was already the second of twelve, see the table in

However, the Commission specified that this standard could only exclude health services that conflicted with widely accepted fundamental principles.[1671] In the view of the Commission the only ethical standards that could influence the inclusion of a health technology in the LEA are those that are defined as fundamental principles in legislative acts or, alternatively, on which there is almost unanimous consensus.[1672] Reference to principles laid down in legislative acts was thus considered to be the only reasonable criterion and also one that was capable of ensuring coherence in the legal system.[1673] It thus seems that the Commission had set out to assess the compliance of new health technologies with the normative framework established by the democratic legislature, rather than their ethical desirability. Hence, despite the terminological ambiguity, the methodological procedure adopted by the 2002 Commission seems in line with the framework of separation of ethics and law adopted in this dissertation.[1674]

As for the most recently established Commission for the Updating of LEA, the legislation only requires it to use health technology assessment procedures in order to assess the inclusion of new technologies in the benefit basket.[1675] This commitment is confirmed by the "Pact for Health 2019-2021" of 27 May 2019, according to which HTA methodologies should be used to assess the impact of new technologies on the healthcare system when annually updating the Essential Levels of Care.[1676]

Commissione nazionale per la definizione e l'aggiornamento dei Livelli essenziali di assistenza in Falcitelli and Langiano, *La remunerazione delle attività sanitarie* (2007) p. 260.

1671 Commissione nazionale per la definizione e l'aggiornamento dei Livelli essenziali di assistenza in Falcitelli and Langiano, *La remunerazione delle attività sanitarie* (2007) p. 254; Arcà and Cislaghi, 'Percorsi metodologici per l'inserimento o l'esclusione di una prestazione dai Livelli essenziali di assistenza' [2006](2) Tendenze nuove p. 97, 102.

1672 Arcà and Cislaghi, 'Percorsi metodologici per l'inserimento o l'esclusione di una prestazione dai Livelli essenziali di assistenza' [2006](2) Tendenze nuove p. 97, 102.

1673 ibid.

1674 Although a legal representation in the commission might be necessary to verify this compliance with the legal framework.

1675 Art. 1(557) Law no. 208/2015, see Antonelli, 'La garanzia dei livelli essenziali di assistenza nei primi 40 anni del Servizio sanitario nazionale: dall'uniformità all'appropriatezza: efficacia non è dimostrabile in base alle evidenze scientifiche' [2018](7) Federalismi p. 1, 20.

1676 Conferenza Stato-Regioni, 'Intesa, ai sensi dell'articolo 8, comma 6, della legge 5 giugno 2003, n. 131 , tra il Governo, le Regioni e le Province autonome di Trento e di Bolzano concernente il Patto per la salute per gli anni 2019-2021' (18.12.2019); see Aperio Bella, 'Tecnologie innovative nel settore salute tra scarsità delle risorse e

This normative benchmark leaves room for the Commission to flesh out its assessment methodology with the criteria it deems necessary. These must, however, be consistent with the legislative framework establishing the requirements of appropriateness, effectiveness and quality of care.

Such standards appear to be fully met in the case of NIPT. Invasive prenatal diagnoses are already considered part of the LEA and compared to them NIPT can be considered to be a more appropriate healthcare technology. The CSS guidelines have also observed that the inclusion of non-invasive diagnoses in the Essential Levels of Care is necessary to ensure compliance with the criterion of appropriateness and in order to prevent the carrying out of risky diagnoses.[1677] It can be anticipated that the Commission will largely draw upon these guidelines when assessing NIPT for inclusion in the benefit basket.

The consideration of 'ethical' issues could only legitimately take place within the scope defined by the former Commission, whereby the crucial factor in assessing the ethics of a health technology is its compliance with normative principles that have been established by the legislature. Therefore the Commission is expected to take into account, for instance, the need for adequate informed consent and counselling, as enshrined in the Constitution under the combination of Articles 2, 13 and 32, as well as in the recent Law no. 219/2017 on informed consent.

C. Non-Invasive Prenatal Testing in England

I. NIPT in the Private Sector

Non-invasive prenatal testing for trisomies 13, 18 and 21 has been available in the United Kingdom since 2012. Its entry onto the UK market was governed by the, then current, Medical Devices Regulations 2002. This legislation gave effect to the European Directives on medical devices and on in vitro diagnostic medical devices[1678] in UK law and regulated the assessment

differenziazione: alla ricerca di un equilibrio difficile' [2020](2) Federalismi p. 245, 260.

1677 Consiglio Superiore di Sanità, Sez. I, 'Screening del DNA fetale non invasivo (NIPT) in sanità pubblica', 9.3.2021, p. 4.

1678 EU Directive 93/42/EEC on medical devices and EU Directive 98/79/EC on in vitro diagnostic medical devices.

procedure for in vitro diagnostic medical devices before notified bodies.[1679] Under these regulations, the market availability of in vitro diagnostic medical devices, such as NIPT, was conditional only on the control of certain essential requirements of quality and safety by the notified bodies.[1680]

Under this regime NIPT has been widely available for purchase in the private sector since 2012, accessible to those patients who could afford to pay for it. Private NIPT providers do not just provide tests to detect trisomies, but also offer to disclose the sex of the foetus.[1681] The tests are either performed through private clinics or obtainable in a 'direct-to-consumer' format, whereby the patient can order the test online and have it performed by a medical practitioner.[1682]

This wide offer of NIPT, accessible through the private sector, raised a number of concerns, especially given the initial lack of its availability in the public sector. Obvious concerns were voiced about potential inequalities arising from the initial high cost of testing in the private sector. This meant that only wealthy patients could afford access to a less invasive test, while less well-off women had to settle for the more invasive and risky tests offered by the public sector.[1683] The main cause for concern, however, was the lack of guarantees on the quality of information offered to pregnant women in the private sector.[1684] Poor information by private providers, often accompanied by misleading statements, affected women's ability to

1679 The Medical Devices Regulations 2002, Reg. 42. The competent authority for implementing medical device legislation and designating notified bodies was and remains the Medicines and Healthcare Products Regulatory Agency (MHRA), an executive agency of the Department of Health and Social Care. For more information, see <https://www.gov.uk/government/organisations/medicines-and-healthcare-products-regulatory-agency> accessed 28.3.2022.

1680 The Medical Devices Regulations 2002, Reg. 34.

1681 Wale, 'Don't Forget the Legal Framework: The Public Provision of Non-invasive Prenatal Testing in England and Wales' (2016) 15(4) Med Law Int p. 203, 205.

1682 Nuffield Council on Bioethics, 'Non-invasive Prenatal Testing: Ethical Issues', London 2017, p. 91.

1683 "This means that there is potential for health inequalities to be created or worsened by the fact that the goods of NIPT are, at the moment, inaccessible to those with less financial means. It might be thought unfair that those who are already better off financially may benefit exclusively from the enhanced choice that NIPT can provide", Nuffield Council on Bioethics, 'Non-invasive Prenatal Testing: Ethical Issues' (London 2017), p. 33.

1684 ibid, pp. 93-ff.

fully understand the procedure and to give their informed consent.[1685] In addition, it was perceived that the private sector would fail to offer the necessary support to women after a positive result, especially when in the form of direct-to-consumer testing.[1686] The lack of adequate follow-up support and counselling was confirmed by the fact that many patients had to seek advice and clarification from NHS medical staff regarding the outcome of privately performed NIPT.[1687]

Evidence that these worries were well founded came when, in 2019, the UK Advertising Standards Authority – an independent organisation regulating advertising practices – issued three rulings declaring the advertisement and information practices of some NIPT providers to be misleading and contrary to the standards developed by the Committee of Advertising Practice. In particular, the marketing material available online exaggerated the accuracy of the test.[1688]

As has been suggested, concerns about health inequalities, misleading information and lack of counselling can, at least partially, be tackled by introducing NIPT into the NHS. Publicly offering these tests free of charge is likely to limit the reach of the private sector.[1689]

1685 "[T]he information and support provided by the private sector may in some cases be affecting the ability of women and couples to make informed choices about NIPT, Nuffield Council on Bioethics, 'Non-invasive Prenatal Testing: Ethical Issues' (London 2017), p. 98.

1686 Nuffield Council on Bioethics, 'Non-invasive Prenatal Testing: Ethical Issues', London 2017, p. 96; Joynson, 'Our concerns about non-invasive prenatal testing (NIPT) in the private healthcare sector' (8.2.2019) <https://www.nuffieldbioethics.org/blog/nipt-private> accessed 23.3.2022.

1687 Jackson, 'Regulating Non-Invasive Prenatal Testing: the view from the UK' [2014] (50) Japanese Journal of Law and Political Science p. 9, 17; Joynson, 'Our concerns about non-invasive prenatal testing (NIPT) in the private healthcare sector', 8.2.2019.

1688 ASA, 'Ruling on The Birth Company: Complaint Ref: A19-564688' (20.11.2019) <https://www.asa.org.uk/rulings/the-birth-company-A19-564688.html> accessed 23.3.2022; ASA, 'Ruling on My Baby Enterprises Ltd: Complaint Ref: A19-564685' (20.11.2019) <https://www.asa.org.uk/rulings/my-baby-enterprises-ltd-A19-564685.html> accessed 23.3.2022; ASA, 'Ruling on Ultrasound Direct Ltd: Complaint Ref: A19-564681' (20.11.2019) <https://www.asa.org.uk/rulings/ultrasound-direct-ltd-A19-564681.html> accessed 23.3.2023.

1689 Brownsword and Wale, 'Testing Times Ahead' (2018) 81(4) Mod Law Rev p. 646, 661.

II. NIPT in the NHS

1. Access to Prenatal Screening and Diagnoses

a Prenatal Screening and Diagnoses in the NHS

Before the introduction of non-invasive prenatal testing for the screening of chromosomal trisomies, the antenatal screening programme offered by the NHS was already quite comprehensive and far-reaching.[1690] All women in the first trimester of pregnancy are offered screening for Down's, Edwards' and Patau's syndrome by means of a 'combined test'. This test takes into account maternal age in combination with the result of ultrasound measurements and the analysis of biochemical markers in the maternal blood.[1691] A 20-week screening scan is also offered that, in addition to chromosomal trisomies, can identify eleven physical malformations including neural tube defects and abdominal wall defects.[1692] In some cases women may have access to a quadruple test in the second trimester of pregnancy to assess the chances of Down's syndrome.[1693]

The provision of this screening programme is in line with NICE's recommendation in its guidance for antenatal care, which advises that all women should be offered screening for chromosomal trisomies in the first trimester.[1694]

1690 Wale, 'Don't Forget the Legal Framework' (2016) 15(4) Med Law Int p. 203, 204–205.

1691 Public Health England, 'Guidance. Down's syndrome, Edwards' syndrome and Patau's syndrome screening pathway requirements specification' (21.6.2021) <https://www.gov.uk/government/publications/downs-syndrome-edwards-syndrome-and-pataus-syndrome-screening-pathway-requirements-specification/downs-syndrome-edwards-syndrome-and-pataus-syndrome-screening-pathway-requirements-specification> accessed 23.3.2022. See Nuffield Council on Bioethics, 'Non-invasive Prenatal Testing: Ethical Issues', London 2017, pp. 7-8.

1692 Public Health England, 'Guidance. 20-week screening scan pathway requirements specification' (21.6.2021) <https://www.gov.uk/government/publications/20-week-screening-scan-pathway-requirements-specification/20-week-screening-scan-pathway-requirements-specification> accessed 23.3.2022.

1693 Details on the difference between these tests can be found in Public Health England, 'Guidance. Screening for Down's syndrome, Edwards' syndrome and Patau's syndrome' (10.12.2021) <https://www.gov.uk/government/publications/fetal-anomaly-screening-programme-handbook/screening-for-downs-syndrome-edwards-syndrome-and-pataus-syndrome--3#quadruple-test> accessed 23.3.2022.

1694 National Institute for Health and Care Excellence, 'Antenatal care: Guideline NG201' (19.8.2021), recommendations no. 1.2.14 and 1.2.15 <https://

If the test result reveals a high possibility that the foetus has a chromosomal trisomy - calculated as a 1 in 150 chance of having an affected foetus - the patient is offered the possibility of confirming this result by invasive diagnosis, i.e. chorionic villus sampling and amniocentesis.[1695] With regard to the uptake of the screening, estimates prior to the introduction of NIPT calculated it at 74 per cent of all women benefiting from NHS services. It was also calculated that, despite the increasing uptake of screenings, the proportion of children born with Down's syndrome had remained fairly constant over the past 25 years.[1696]

In order to receive NHS funding, screening tests must be of a certain medical relevance. That is, they should provide information that may be relevant either to possible prenatal treatment, though this is not available in the case of chromosomal trisomies, or to enable the woman to consider terminating the pregnancy. The last option only comes into question in cases covered by the provisions of the Abortion Act 1967 and in particular section 1(1)(d), according to which the patient may request an abortion if two registered medical practitioners are of the opinion that there is a substantial risk the child would suffer from serious physical or mental conditions.[1697] In this respect, the aim of including these screening tests within NHS

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www.nice.org.uk/guidance/ng201/chapter/Recommendations> accessed 23.3.2022 This guidance updates and replaces previous NICE guidance CG62 that recommended that the 'combined test' to screen for Down's syndrome should be offered to all pregnant women should be offered screening for Down's syndrome and that women should understand that it is their choice to embark on this procedure, National Institute for Health and Care Excellence, 'Antenatal care for uncomplicated pregnancies: Clinical guideline CG62' (4.2.2019) <https://www.nice.org.uk/guidance/cg62> accessed 23.3.2022. On this policy decision, see Scott, *Choosing Between Possible Lives* (2007) p. 177.

1695 Nuffield Council on Bioethics, 'Non-invasive Prenatal Testing: Ethical Issues', London 2017, p. 8; Public Health England, 'Guidance. Prenatal diagnosis' (10.12.2021) <https://www.gov.uk/government/publications/fetal-anomaly-screening-programme-handbook/prenatal-diagnosis> accessed 23.3.2022.

1696 "The proportion of women having a termination after a diagnosis has remained steady, ranging from 89 to 95 per cent between 1989 and 2012, meaning that the actual number of terminations has increased. However, the number of live births of babies with Down's syndrome has remained fairly constant. This is likely to be due to an increased incidence of Down's syndrome in fetuses caused by an increase in the average age of women at delivery", Nuffield Council on Bioethics, 'Non-invasive Prenatal Testing: Ethical Issues', London 2017, p. 9.

1697 "For any new screening or testing to become part of the standard of care in England, this must ultimately have some connection with the terms of the disability ground of the Abortion Act", Scott, *Choosing Between Possible Lives* (2007) p. 193.

care is to contribute to women's reproductive autonomy, enabling them to make informed choices about their pregnancies. This is both in terms of continuing the pregnancy with the benefit of additional information about the special needs of the developing foetus and also in terms of opting for an abortion procedure.

b Autonomy and Informed Consent

To achieve the overarching goal of facilitating women's reproductive autonomy it is essential for the NHS to guarantee that screening procedures are offered in a way that is compatible with the patients' fully informed consent. In order to achieve an improvement in reproductive autonomy the information provided to the patient about screening for chromosomal trisomies must meet certain requirements. First of all, it must be clear that screening is in all its stages entirely voluntary.[1698] Healthcare professionals must refrain from creating any pressure that would make the woman feel obliged to accept the offer of screening.[1699] In addition, the given information should include details about the conditions for which screening is performed and about the quality of life of children born with chromosomal trisomies.[1700]

The provision of comprehensive and detailed information about a pregnant woman's diagnostic and treatment options is also a common law requirement, the violation of which can amount to clinical negligence. Obligations to obtain the patient's informed consent have indeed become more stringent following the 2015 landmark decision of the UK Supreme Court in the case of *Montgomery v Lanarkshire Health Board*.[1701] The case

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See also Wale, 'Don't Forget the Legal Framework' (2016) 15(4) Med Law Int p. 203, 211.

1698 Scott, *Choosing Between Possible Lives* (2007) p. 146.

1699 UK Human Genetics Commission, 'Making Babies' (2006) 11(1) Jahrbuch für Wissenschaft und Ethik p. 485, para. 20; Scott, *Choosing Between Possible Lives* (2007) p. 146.

1700 „To give valid consent, a woman must also be informed about the nature of any screening or testing. Arguably 'nature' includes purpose (rather than just the physical nature of a test, eg, the mechanisms of an ultrasound scan or the taking of blood) and in this context 'purpose' should include information about the condition that is the subject of screening.", Scott, *Choosing Between Possible Lives* (2007) p. 149.

1701 *Montgomery v Lanarkshire Health Board* [2015] UKSC 11 (11 March 2015).

concerned a diabetic woman with a high-risk pregnancy whom the doctor had failed to inform about the possible negative consequences of a vaginal delivery for a patient in her condition and about the possibility of alternatively requiring a caesarean section. As a result of the attempt to perform a natural birth, the baby was born with severe disabilities. Whereas, according to previous case law, the support of a responsible body of medical opinion that withstood logical scrutiny was capable of excluding negligence,[1702] the Supreme Court in *Montgomery* overturned this professional standard by according a greater significance to the need to respect the patient's autonomy. As Lady Hale maintained: patient autonomy is an important feature of a person's physical and psychiatric integrity.[1703] The judgment recognised that patients have a right to be given more comprehensive information, for they are "now widely regarded as persons holding rights, rather than as the passive recipients of the care of the medical profession".[1704] This decision thus clearly marks the final realisation of the paradigm shift from medical paternalism to patient rights.[1705] As Lady Hale pointed out, this is particularly true when the doctor's judgment goes beyond a purely medical one and takes on an ethical connotation.[1706] In the *Montgomery* case, for example, it was the idea that vaginal delivery is in some way morally preferable to a caesarean section. [1707] In these circumstances patients are all the more entitled to decide according to their own values.[1708]

1702 According to the so-called *Bolam* test, as developed in the case of *Bolam v Friern Hospital Management Committee* [1957] 1 WLR 582 (26 February 1957).

1703 *Montgomery v Lanarkshire Health Board* [2015] UKSC 11 (11 March 2015), para. 108.

1704 *Montgomery v Lanarkshire Health Board* [2015] UKSC 11 (11 March 2015), para 75.

1705 "Without doubt, the headline story in Montgomery is that the doctor / patient relationship is now predicated on the rights paradigm rather than ethical paradigms that prioritise professional duties or paternalistic responsibilities or that centre on maximising utility or minimising distress", Brownsword and Wale, 'The Development of Non-Invasive Prenatal Testing: Some Legal and Ethical Questions' [2016] (24) JRE p. 31, 41.

1706 *Montgomery v Lanarkshire Health Board* [2015] UKSC 11 (11 March 2015), para 114. See Sutherland Qc, 'The Right of Patients to Make Autonomous Choices: Montgomery v Lanarkshire Health Board: A Landmark Decision on Information Disclosure to Patients in the UK' (2021) 32(7) Int Urogynecol J p. 2005, 2007.

1707 *Montgomery v Lanarkshire Health Board* [2015] UKSC 11 (11 March 2015), para. 114.

1708 *Montgomery v Lanarkshire Health Board* [2015] UKSC 11 (11 March 2015), para. 115.

The court indicated that the scope of the information to be given to the patient can be inferred by means of a 'materiality test'. The doctor must thereby disclose all diagnostic and therapeutic possibilities that a 'reasonable person' or that particular patient (where the doctor should reasonably have been aware of the relevant particularities) might consider relevant.[1709]

The implication for prenatal screening and diagnosis is, on the one hand, that the doctor must inform the woman of any tests that may disclose a risk to which she "would be likely to attach significance".[1710] On the other hand, respect for the value judgements of the particular patient must be maintained and this implies not only that the woman also has a right not to know, but also that she must be made aware of all the circumstances necessary to make an informed choice.[1711] Sometimes in clinical practice the extent of the information provided is insufficient when it comes to non-invasive methods of screening, such as ultrasound.[1712] This stems from the fact that there is no risk to the foetus or the patient in performing the test. However, based on *Montgomery*, the information to be given to the patient goes beyond the possible risks involved in the testing and encompasses the consequences of screening, the accuracy of the results and clarifications on the conditions that can be detected.

As argued by the Nuffield Council of Bioethics (NCOB) in 2017, a possible consequence of *Montgomery v Lanarkshire Health Board* was that, before NIPT was finally offered by the NHS, doctors were required to inform women of its availability in the private sector as an alternative.[1713]

1709 *Montgomery v Lanarkshire Health Board* [2015] UKSC 11 (11 March 2015), para. 87.

1710 Montgomery v Lanarkshire Health Board [2015] UKSC 11 (11 March 2015), On the 'materiality test' in prenatal screening, see Scott, *Choosing Between Possible Lives* (2007) pp. 173- 174.

1711 Brownsword and Wale, 'Testing Times Ahead' (2018) 81(4) Mod Law Rev p. 646, 651.

1712 See Ravitsky, 'The Shifting Landscape of Prenatal Testing: Between Reproductive Autonomy and Public Health' (2017) 47(Suppl 3) Hastings Cent Rep S34-S40, S35, who claims that "[t]he informed-consent process for ultrasound has been completely abandoned".

1713 Nuffield Council on Bioethics, 'Non-invasive Prenatal Testing: Ethical Issues', London 2017, p. 41. The obligation to inform the patients is all the more valid in case of inclusion of NIPT in the NHS care, see Brownsword and Wale, 'The Development of Non-Invasive Prenatal Testing: Some Legal and Ethical Questions' [2016](24) JRE p. 31, 42: "after *Montgomery*, we suggest that it is reasonable to assume that, at all stages of a pregnancy, whether in the ante-natal screening clinic or in the delivery room, a woman has a right to be informed about the options that are available to her. It follows that, once NIPT is embedded in the screening

2. Evaluation Procedure before the UK National Screening Committee

a The UK National Screening Committee's Recommendation

Following its introduction into the private sector, an evaluation of NIPT for Down's, Edward's and Patau's syndrome was undertaken by the UK National Screening Committee (UK NSC). This is an independent advisory body responsible for assessing all aspects of screening programmes and making recommendations to health ministers and to the NHS across the UK.[1714]

In England the Secretary of State is responsible for defining screening programmes based on the recommendations of the UK NSC, while NHS England[1715] provides the commissioning and delivery of the service in exercise of the public health functions delegated to it according to Section 7A of the National Health Service Act 2006.[1716]

The value of the decisions of the UK National Screening Committee is recognised by the NHS Constitution for England, which states that the NHS is committed to providing the population with the screening

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pathway, pregnant women will have a right to know about the availability of the test, and to be informed about the risks and consequences of having the test".

1714 For information on the UK NSC, see <https://www.gov.uk/government/organisations/uk-national-screening-committee/about> accessed 23.3.2022. See also Mauthoor, 'Five things you should know about the UK NSC' (7.6.2021) <https://nationalscreening.blog.gov.uk/2021/06/07/five-things-you-should-know-about-the-uk-nsc/> accessed 23.3.2022.

1715 Since end of 2021, previously this task was entrusted to Public Health England. The passing of delegated functions with regard to screening to NHS England occurred in October 2021 through a letter of the Department of Health and Social Care to NHS England, see Department of Health and Social Care, 'NHS public health functions (section 7A) agreement 2021 to 2022: letter from DHSC to NHSE' (18.11.2021) <https://www.gov.uk/government/publications/public-health-commissioning-in-the-nhs-2021-to-2022/nhs-public-health-functions-section-7a-agreement-2021-to-2022-letter-from-dhsc-to-nhse> accessed 23.3.2022

1716 National Health Service Act 2006 sec. 7A. The NHS fetal anomaly screening programme is included in the public health functions delegated by the Secretary of State to NHS England according to this section, see Department of Health and Social Care, 'Annex: public health functions (section 7A) agreement 2020 to 2021 – services to be provided' (26.10.2020) <https://www.gov.uk/government/publications/public-health-commissioning-in-the-nhs-2020-to-2021/annex-public-health-functions-section-7a-agreement-2020-to-2021-services-to-be-provided> accessed 23.3.2022

programmes recommended by the Committee.[1717] Once approved by the health ministers, screening programmes that have been recommended by the UK NSC are publicly funded and offered free of charge to patients.[1718]

In order to best inform its recommendation on NIPT the UK NSC first gathered scientific evidence by analysing the medical literature and the results of clinical trials. Moreover, in order to gain an insight into the impact of the introduction of NIPT into clinical practice in an NHS setting, the Committee supported the initiation of the RAPID study, funded by the National Institute for Health Research, evaluating the use of NIPT for Down's syndrome in several NHS maternity units.[1719] This study implemented a sort of 'coverage with evidence development' scheme in that it allowed the UK NSC to obtain more information about the accuracy, cost and effectiveness of screening for Down's syndrome with NIPT, while the costs of the test could be publicly covered for all patients recruited as study participants. The key aim of the RAPID study was to obtain data necessary to evaluate, *inter alia*, the accuracy of NIPT in low-risk pregnancies, its cost-effectiveness and uptake, as well as the possibility of maintaining informed choice in accepting or declining testing.[1720]

The study concluded that implementing NIPT in the NHS screening programme for Down's syndrome could "improve quality of care, choices for women, and overall performance within the current budget".[1721] This outcome can be reached by offering NIPT as a contingent test, depending on the results of the first screening. It was calculated that the accuracy of the test would only be guaranteed if it was conducted within a population for which the initial screening had revealed a chance of at least 1 in 150 of having a foetus with Down's syndrome.[1722]

1717 Department of Health and Social Care, 'The NHS Constitution for England', 1.1.2021

1718 See Ravitsky and others, 'The Emergence and Global Spread of Noninvasive Prenatal Testing' (2021) 22(1) Annu Rev Genom Hum Genet p. 309, 324.

1719 Hill and others, 'Evaluation of Non-invasive Prenatal Testing (NIPT) for Aneuploidy in an NHS Setting: A Reliable Accurate Prenatal Non-invasive Diagnosis (RAPID) Protocol' (2014) 14(229) BMC Pregnancy Childbirth p. 1, 11.

1720 ibid, p. 3.

1721 Chitty and others, 'Uptake, Outcomes, and Costs of Implementing Non-invasive Prenatal Testing for Down's Syndrome into NHS Maternity Care: Prospective Cohort Study in Eight Diverse Maternity Units' (2016) 354(i3426) BMJ p. 1

1722 ibid, p. 9.

The study also revealed that about one third of women with a positive NIPT result decided to continue with the pregnancy.[1723] The non-invasive nature of the test also allows it to be used by those women who would like to have more information in order to prepare for giving birth to a child with a chromosomal aneuploidy. The findings of the study thus suggested that the number of children born with Down's syndrome may not vary significantly with the introduction of NIPT in the public sector.[1724] Moreover, it was argued that guaranteeing a high level of informed consent is both necessary and achievable.[1725]

In terms of costs it was estimated that the introduction of NIPT into NHS maternal care would be cost-neutral or even result in a slight reduction in expenses due to the fact that many invasive procedures would be avoided.[1726]

In mid-2015 the RAPID study team reported the evidence and its positive assessment of NIPT to the UK NSC.[1727] On this basis, and aware of the different opinions on the implementation of the test in the NHS, the UK NSC decided to launch a three-month public consultation at its June

1723 ibid, p. 10. In another study, termination of pregnancy was chosen by 74% of the patients, see Gil and others, 'Clinical Implementation of Routine Screening for Fetal Trisomies in the UK NHS: Cell-free DNA Test Contingent on Results from First-trimester Combined Test' (2016) 47(1) Ultrasound Obstet Gynecol p. 45, 51.

1724 Chitty and others, 'Uptake, Outcomes, and Costs of Implementing Non-invasive Prenatal Testing for Down's Syndrome into NHS Maternity Care' (2016) 354(i3426) BMJ p. 1, 10. While "[t]he overall proportion of terminations of pregnancy following a diagnosis of Down's syndrome is likely to fall, […] the number of terminations is likely to increase", Nuffield Council on Bioethics, 'Non-invasive Prenatal Testing: Ethical Issues', London 2017, 51.

1725 On this point see Ravitsky and others, 'The Emergence and Global Spread of Noninvasive Prenatal Testing' (2021) 22(1) Annu Rev Genom Hum Genet p. 309, p. 324.

1726 Chitty and others, 'Uptake, Outcomes, and Costs of Implementing Non-invasive Prenatal Testing for Down's Syndrome into NHS Maternity Care' (2016) 354(i3426) BMJ p. 1, 11; Mackie, 'Addition of non-invasive test to screening for Down's syndrome, Edward's syndrome, Patau's syndrome' (3.11.2016) <https://nationalscreening.blog.gov.uk/2016/11/03/addition-of-non-invasive-test-to-improve-screening-for-pregnant-women/> accessed 23.3.2022; Nuffield Council on Bioethics, 'Non-invasive Prenatal Testing: Ethical Issues', London 2017, p. 39.

1727 Chitty and others, 'Uptake, Outcomes, and Costs of Implementing Non-invasive Prenatal Testing for Down's Syndrome into NHS Maternity Care' (2016) 354(i3426) BMJ p. 1, 11; Wale, 'Don't Forget the Legal Framework' (2016) 15(4) Med Law Int p. 203, 206.

2015 meeting, before issuing its final recommendation.[1728] The consultation sought reactions to the proposal to publicly fund NIPT only for women who were found to be at a higher risk after the combined test.[1729]

The 30 stakeholders who responded to the consultation offered the committee a variety of voices and perspectives. [1730] However, the majority of them reacted positively to the proposal and some argued that the risk threshold for accessing the test should be reduced.[1731] Respondents opposed to the inclusion of NIPT in the public service were mainly those who were fundamentally against all prenatal screening for chromosomal aneuploidies that could lead to abortion.[1732]

Based on the research and evidence gathered in the evaluation process, the UK NSC decided in January 2016 to recommend an evaluative implementation of NIPT within the existing NHS Fetal Anomaly Screening Programme.[1733] The recommendation to include NIPT in an initially cautious and controlled manner showed a pragmatic approach while at the same time taking into account ethical issues and the relatively new nature of the test.[1734] The UK NSC hoped that the evaluative roll out in the NHS would provide a better understanding of the impact of publicly funded

1728 UK National Screening Committee, 'Note of the meeting held on the 18 June 2015' <https://www.gov.uk/government/publications/uk-nsc-meeting-june-2015> accessed 23.3.2022

1729 ibid.

1730 Marshall, 'Evidence update: consultation on non-invasive prenatal testing and latest UK NSC recommendations' (13.8.2022) <https://nationalscreening.blog.gov.uk/2015/08/13/evidence-update-new-consultation-on-non-invasive-prenatal-testing-and-latest-uk-nsc-recommendations/>.

1731 UK National Screening Committee, 'Note of the meeting held on the 19 November 2015' <https://www.gov.uk/government/publications/uk-nsc-meeting-november-2015> accessed 23.3.2022; Ravitsky and others, 'The Emergence and Global Spread of Noninvasive Prenatal Testing' (2021) 22(1) Annu Rev Genom Hum Genet p. 309, 325.

1732 UK National Screening Committee, 'Note of the meeting held on the 19 November 2015'; Ravitsky and others, 'The Emergence and Global Spread of Noninvasive Prenatal Testing' (2021) 22(1) Annu Rev Genom Hum Genet p. 309, 325.

1733 UK National Screening Committee, 'UK NSC non-invasive prenatal testing (NIPT) recommendation' (01.2016) <https://legacyscreening.phe.org.uk/policydb_download.php?doc=602> accessed 23.3.2022.

1734 UK National Screening Committee, 'Note of the meeting held on the 19 November 2015'.

NIPT on the reproductive autonomy of pregnant women before its full and permanent implementation.[1735]

The UK NSC recommendation confirmed the option for contingent use of NIPT, i.e. dependent on the results of initial screening. It was recommended that women should be offered the usual combined ultrasound and blood test or other non-invasive screening in the first trimester and that NIPT for trisomy 21, 13 and 18 should only be offered to women who exceed the risk threshold of 1 in 150.[1736] Among these, women who received a positive NIPT result would be advised to seek amniocentesis or chorionic villus sampling, whereas there would be no need for an invasive test in the case of a negative NIPT finding.[1737] Accordingly, NIPT is not offered as a standard test to all women, but still helps to avoid the majority of invasive procedures with a risk of miscarriage.[1738]

b Reactions to the UK NSC's Assessment

Following the UK NSC's recommendation extensive media coverage addressed the effects of offering NIPT in the public sector on people with disabilities. Public debate was especially prompted by a highly successful BBC documentary, presented in October 2016 by actor Sally Phillips,

1735 Mackie, 'Addition of non-invasive test to screening for Down's syndrome, Edward's syndrome, Patau's syndrome', 3.11.2016; Nuffield Council on Bioethics, 'Non-invasive Prenatal Testing: Ethical Issues', London 2017, p. 37. According to Brownsword and Wale "the 'piloting' of NIPT within the NHS Fetal Anomaly Screening Programme, leaves its status somewhere between 'research' and 'implementation'", Brownsword and Wale, 'Testing Times Ahead' (2018) 81(4) Mod Law Rev p. 646, 672.

1736 "Covering NIPT for all pregnancies was not deemed cost effective in terms of anticipated savings to the health care system (compared with the current program) with respect to a reduction in the number of invasive tests and the anticipated number of Down syndrome diagnoses during pregnancy", Ravitsky, 'The Shifting Landscape of Prenatal Testing' (2017) 47(Suppl 3) Hastings Cent Rep S34-S40, S37. See also Ravitsky and others, 'The Emergence and Global Spread of Noninvasive Prenatal Testing' (2021) 22(1) Annu Rev Genom Hum Genet p. 309, 324.

1737 See Brownsword and Wale, 'Testing Times Ahead' (2018) 81(4) Mod Law Rev p. 646, 647–648.

1738 Wale, 'Regulating Disruptive Technology and Informational Interests in the Arena of Reproductive Tests' (2019) 3(1) Journal of Information Rights, Policy and Practice p. 1, 3.

who has a child with Down's syndrome.[1739] The documentary argued that women who are offered screening are often given misleading and biased information on the condition.[1740] Hence, the provision of cost-free NHS testing would lead to an increase in abortions of foetuses with chromosomal aneuploidies and an overall decrease in the births of children with Down's syndrome.[1741] In the same year a Down's syndrome advocacy group launched a petition and awareness campaign under the slogan 'Don't Screen Us Out'.[1742] The campaign argued that the introduction of NIPT into the NHS setting would give pregnant women the impression that screening for trisomy 21 is encouraged and hard to turn down.[1743] It was alleged that, eventually, the public funding of NIPT would result in a greater routinisation of screening, poor information for pregnant women and in the stigmatisation of people with disabilities.[1744]

These concerns were expressed in a letter to the Department of Health in which the government was accused of failing to properly consult the community of people with Down's syndrome.[1745] A parliamentary motion signed by thirty-four MPs of different political parties joined in support of the campaign and asked the government to postpone the implementation

1739 Phillips and Richards, 'A World Without Down's Syndrome' (First Broadcast 5.10.2016) BBC <https://www.bbc.co.uk/programmes/b07ycbj5> accessed 6.4.2022; Burch, 'A world without Down's syndrome?: Online resistance on Twitter: #worldwithoutdowns and #justaboutcoping' (2017) 32(7) Disability & Society p. 1085. See Nuffield Council on Bioethics, 'Non-invasive Prenatal Testing: Ethical Issues', London 2017, pp. 14-15.

1740 Perrot and Horn, 'The Ethical Landscape(s) of Non-invasive Prenatal Testing in England, France and Germany' (2022) 30 Eur J Hum Genet p. 676, 678.

1741 Although the RAPID study had suggested that "Down's syndrome live birth rates may not change significantly", see Chitty and others, 'Uptake, Outcomes, and Costs of Implementing Non-invasive Prenatal Testing for Down's Syndrome into NHS Maternity Care' (2016) 354(i3426) BMJ p. 1, 11.

1742 Ravitsky, 'The Shifting Landscape of Prenatal Testing' (2017) 47(Suppl 3) Hastings Cent Rep S34-S40, S37; Perrot and Horn, 'The Ethical Landscape(s) of Non-invasive Prenatal Testing in England, France and Germany' (2022) 30 Eur J Hum Genet p. 676, 678.

1743 Ravitsky, 'The Shifting Landscape of Prenatal Testing' (2017) 47(Suppl 3) Hastings Cent Rep S34-S40, S37.

1744 Brownsword and Wale, 'The Development of Non-Invasive Prenatal Testing: Some Legal and Ethical Questions' [2016](24) JRE p. 31, 32.

1745 As reported by Ravitsky, 'The Shifting Landscape of Prenatal Testing' (2017) 47(Suppl 3) Hastings Cent Rep S34-S40, S37; Iacobucci, 'Non-invasive Prenatal Testing: Public and Doctors Should be Consulted, says BMA' (2018) 362(k2916) BMJ p. 1.

of NIPT in the NHS until possible discriminatory effects on people with Down's syndrome and their families had been investigated and prevented.[1746] The British Medical Association also advocated wider consultation on the views of the public and medical profession.[1747]

Fears of increased discrimination against people with Down's syndrome have been intensified by a controversy involving the United Kingdom's Royal College of Obstetricians and Gynaecologists (RCOG). The RCOG was accused of suggesting, in its response to the public consultation conducted by the UK NSC, that the lifetime costs of caring for a child with Down's syndrome should have been included in the economic cost-effectiveness analysis.[1748] However, on the one hand, the RCOG argued that this was a misunderstanding of their statement[1749] and, on the other hand, this perspective had not been embraced by the UK NSC in its recommendation.

Although the voices of advocacy groups were prominent in the public debate, the inclusion of NIPT in the existing NHS Fetal Anomaly Screening Programme enjoyed widespread public support.[1750] Most members of society, including some belonging to the Down's syndrome community,[1751] supported women's reproductive autonomy and their right to obtain comprehensive information about the health of the foetus.[1752] It was emphasised that NIPT had the advantage of reducing the invasiveness and risks of miscarriage associated with the existing screening programme. This was

1746 UK Parliament, 'Early Day Motion 44: Down's Syndrome, Don't Screen Us Out Campaign' (19.5.2016) <https://edm.parliament.uk/early-day-motion/49295/downs-syndrome-dont-screen-us-out-campaign> accessed 23.3.2022. See Ravitsky, 'The Shifting Landscape of Prenatal Testing' (2017) 47(Suppl 3) Hastings Cent Rep S34-S40, S37.

1747 Iacobucci, 'Non-invasive Prenatal Testing' (2018) 362(k2916) BMJ p. 1.

1748 Ravitsky, 'The Shifting Landscape of Prenatal Testing' (2017) 47(Suppl 3) Hastings Cent Rep S34-S40, S37-S38.

1749 Wise, 'The End of Down's Syndrome?' (2016) 355(i5344) BMJ p. 1, 2.

1750 Ravitsky, 'The Shifting Landscape of Prenatal Testing' (2017) 47(Suppl 3) Hastings Cent Rep S34-S40, S37.

1751 Nuffield Council on Bioethics, 'Non-invasive Prenatal Testing: Ethical Issues', London 2017, p. 54; Perrot and Horn, 'Preserving Women's Reproductive Autonomy While Promoting the Rights of People with Disabilities?' [2022](0) J Med Ethics p. 1, 2.

1752 Perrot and Horn, 'The Ethical Landscape(s) of Non-invasive Prenatal Testing in England, France and Germany' (2022) 30 Eur J Hum Genet p. 676, 678.

seen as a positive development and an "if not entirely unproblematic, at least relatively uncontroversial"[1753] innovation.

Also the report on the ethical issues of NIPT, issued by the Nuffield Council of Bioethics after extensive public consultation, was critical but ultimately supportive of the UK NSC recommendations.[1754]

Some legal scholars have even described the approach of the UK NSC and the Nuffield Council of Bioethics as relatively conservative[1755] and have pointed out the desirable aspects of including non-invasive screening technologies in NHS care.[1756] Indeed, emphasis has been placed on the fact that publicly funded NIPT increases the quality of the health service. Firstly, it reduces the inequality between wealthy couples, who can afford safer tests in the private sector, and those who lack financial means.[1757] In addition, NIPT limits the overall amount of invasive procedures required. Finally, it improves women's reproductive health and physical and psychological well-being, both by enabling them to decide for an abortion and in terms of preparedness for the birth of a child with chromosomal aneuploidies.[1758]

The effective improvement of the quality of the health service obviously presupposes high standards of information and counselling, as well as the guarantee of fully informed consent. It is important to ensure that women do not feel obliged to participate in the screening programme and that they are not misled as to the implications of having a child with a chromosomal trisomy. Adequate NHS screening programmes therefore also include education and training for health professionals.[1759]

In this respect, offering NIPT within the public sector has the advantage of allowing control over the quantity and quality of information given to

1753 Brownsword and Wale, 'The Development of Non-Invasive Prenatal Testing: Some Legal and Ethical Questions' [2016](24) JRE p. 31.

1754 Details of this report are outlined below.

1755 Brownsword and Wale, 'Testing Times Ahead' (2018) 81(4) Mod Law Rev p. 646, 672.

1756 Scott, *Choosing Between Possible Lives* (2007) p. 176.

1757 Wale, 'Don't Forget the Legal Framework' (2016) 15(4) Med Law Int p. 203, 208.

1758 Scott, *Choosing Between Possible Lives* (2007) 176; Wale, 'Don't Forget the Legal Framework' (2016) 15(4) Med Law Int p. 203, 208; Brownsword and Wale, 'The Development of Non-Invasive Prenatal Testing: Some Legal and Ethical Questions' [2016](24) JRE p. 31, 35.

1759 Ravitsky and others, 'The Emergence and Global Spread of Noninvasive Prenatal Testing' (2021) 22(1) Annu Rev Genom Hum Genet p. 309, 324.

patients.[1760] While the private sector has an interest in offering as much testing as possible and persuading women into believing that the tests are entirely accurate, implementation in the public sector has the potential to ensure that information is neutral and aimed at the full realisation of women's reproductive autonomy.

c Evaluative Implementation of NIPT in the NHS

In November 2016, the government announced that it would follow the recommendations of the UK NSC and offer NIPT for trisomies 21, 18 and 13 under an evaluative roll out for all women found to be at high risk after initial screening.[1761] The inclusion of NIPT in the NHS fetal anomaly screening programme was due to begin in late 2018. The implementation of the screening programme was entrusted to Public Health England, a former executive agency of the Department of Health. The evaluation period was planned to last three years during which the effects of publicly offering NIPT could be monitored and the screening programme modified as necessary.[1762]

During the preparation of the evaluative roll out the possibility of a further consultation of advocacy groups and stakeholders was raised. In a parliamentary question the Secretary of State for Health and Social Care was asked if the government would "consider conducting a consultation on

1760 Brownsword and Wale, 'Testing Times Ahead' (2018) 81(4) Mod Law Rev p. 646, 648 and 660.

1761 Department of Health and Social Care, 'News story. Safer screening test for pregnant women: New non-invasive prenatal test for Down's, Edwards' and Patau's syndromes, which is safer for women and their babies.' (02.11.2016) <https://www.gov.uk/government/news/safer-screening-test-for-pregnant-women> accessed 23.3.2022; Nuffield Council on Bioethics, 'Non-invasive Prenatal Testing: Ethical Issues. Review of Activities Since Publication' (November 2018), p. 5 <https://www.nuffieldbioethics.org/assets/pdfs/Nuffield-Council-NIPT-review-of-activites.pdf> accessed 23.3.2022.

1762 "NIPT will be introduced as an 'evaluative roll out'. This means we will be able to monitor how the introduction of NIPT is working at each stage of the roll out and make any changes to the pathway and screening processes quickly and effectively", McHugh, 'NIPT procurement and launch update' (28.1.2021) <https://phescreening.blog.gov.uk/2021/01/28/nipt-procurement-and-launch-update/> accessed 22.3.2023.

the ethical implications of non-invasive prenatal testing".[1763] However, the government considered that the extensive work that the UK NSC and the Nuffield Council of Bioethics had already done, in terms of consultation and in terms of assessing the ethical issues, was sufficient. It was rather necessary to review and assess the test in practice.[1764]

While the evaluative introduction of NIPT was promptly implemented in Wales in April 2018,[1765] in Scotland and England there were some delays due to procurement issues.[1766] NIPT was finally implemented in the NHS fetal anomaly screening programme[1767] from 1 June 2021 in most parts of England[1768] and then extended to all maternity care units in July 2021. [1769]

The information booklet 'Screening tests for you and your baby' to be distributed to pregnant women in NHS care has also been updated by the UK NSC to include NIPT. The leaflet contains detailed information on the testing procedure, on each condition screened for and on the voluntariness

1763 Parliamentary question posed by Lavery, 'Pregnancy: Screening. Question for Department of Health and Social Care: UIN 285277 (Answer: Caroline Dinenage)' (2.9.2019) <https://questions-statements.parliament.uk/written-questions/detail/2019-09-02/285277#> accessed 23.3.2022.

1764 ibid.

1765 Public Health Wales, 'New screening for pregnant women to be offered in Wales' <http://www.wales.nhs.uk/news/48260> accessed 23.3.2022.

1766 See parliamentary question posed by Morris, 'Pregnancy: Screening. Question for Department of Health and Social Care: UIN 251394 (Answer: Selma Kennedy)' (7.5.2019) <https://questions-statements.parliament.uk/written-questions/detail/2019-05-07/251394> accessed 23.3.2022. See also McHugh, 'NIPT procurement and launch update', 28.1.2021.

1767 Public Health England, 'Guidance. Screening for Down's syndrome, Edwards' syndrome and Patau's syndrome: NIPT' (23.9.2021) <https://www.gov.uk/government/publications/screening-for-downs-syndrome-edwards-syndrome-and-pataus-syndrome-non-invasive-prenatal-testing-nipt/screening-for-downs-syndrome-edwards-syndrome-and-pataus-syndrome-nipt> accessed 23.3.2022

1768 Mackie, 'NIPT to be evaluated as a new part of NHS screening pathway for Down's syndrome, Edwards' syndrome and Patau's syndrome' (1.6.2021) <https://phescreening.blog.gov.uk/2021/06/01/nipt-to-be-evaluated-as-a-new-part-of-nhs-screening-pathway-for-downs-syndrome-edwards-syndrome-and-pataus-syndrome/> accessed 23.3.2022.

1769 Permalloo, 'NIPT rolls out to all areas of England as part of the existing NHS screening pathway for Down's syndrome, Edwards' syndrome and Patau's syndrome' (1.7.2021) <https://phescreening.blog.gov.uk/2021/07/01/nipt-rolls-out-to-all-areas-of-england-as-part-of-the-existing-nhs-screening-pathway-for-downs-syndrome-edwards-syndrome-and-pataus-syndrome/> accessed 23.3.2022.

of the participation in the screening programme. [1770] This information has been updated also with the involvement of stakeholders such as associations representing people with disabilities.[1771]

3. Ethical Considerations in the Assessment Procedure of NIPT

a The Nuffield Council of Bioethics' Report on NIPT

Following the recommendations of the UK NSC the Nuffield Council of Bioethics contributed to the debate with the publication of a report on the ethical issues surrounding NIPT. The document sought to consider the ethical and legal implications of NIPT's use in both the private sector and the NHS and to share insights with decision-makers and stakeholders.[1772] The aim was not primarily to provide advice for the government but rather to investigate and illustrate the various ethical viewpoints voiced across society in order to better prepare the ground for informed public participation in the debate.

In this respect the Council's fundamental approach differs, at least in part, from that of bioethics committees in other European countries. Firstly, the NCOB is a non-governmental organisation, which was established independently by a charitable foundation.[1773] Although it has no democratic legitimacy[1774] it has established itself as a *de facto* national ethics committee.

1770 Public Health England, 'Guidance. Screening tests for you and your baby' (3.5.2019) <https://www.gov.uk/government/publications/screening-tests-for-you-and-your-baby> accessed 23.3.2022.

1771 As outlined in the intervention by Dr Elizabeth Corcoran, Chair of the Down's Syndrome Research Foundation at the Conference Prenatal Testing, Disability, and the Ethical Society, 'Reflections Following Crowter' (4.3.2022) <https://www.law.ox.ac.uk/events/prenatal-testing-disability-and-ethical-society-reflections-following-crowter> accessed 23.3.2022.

1772 Nuffield Council on Bioethics, 'Non-invasive Prenatal Testing: Ethical Issues', London 2017, p. x: "[t]he terms of reference of the Working Group on non-invasive prenatal testing (NIPT) were: 1 to consider the ethical, legal and regulatory implications of recent and potential future scientific developments in NIPT, with regard to its use in both NHS and commercial services, including for whole genome/exome sequencing; 2 to engage a range of people and organisations in the consideration of these questions; 3 to report and disseminate findings and recommendations amongst key decision-makers and other stakeholders".

1773 Montgomery in Palazzani, *Role and Functions of Bioethics Committees* (2014).

1774 Montgomery, 'Bioethics after Brexit: An Opportunity to Rationalize Bioethics Governance in the United Kingdom' (2018) 18(2-3) Med Law Int p. 135, 150–151.

The reputation it has acquired guarantees a "tacit acceptance" of its authority, which is "consistent with traditional British political pragmatism".[1775] Secondly, although its reports usually include some recommendations for the decision-makers, these are never their main purpose.[1776] The aim of the Nuffield Council of Bioethics is rather to prepare a basis for adequately informed public discussion on new developments in science and healthcare.[1777] For this purpose the Council sets up a working group for each medical innovation with the task of gathering and systematising different ethical approaches and scientific evidence. The focus is on maintaining broad inclusiveness by ensuring that all opinions can initially be given a voice and only then be put to a test of rigorousness and reasonableness.[1778] Hence, the Council does not strive to establish a definite and consistent ethical paradigm across its various reports.[1779] The legitimacy of the Council's documents is not based on the adoption of certain substantive principles, but rather on compliance with criteria of procedural legitimacy, including gathering evidence, bringing together members with expertise in different areas, conducting public consultations, listening to all sides and applying reasonableness standards.[1780]

These procedural principles were also applied in drafting the report on the ethical issues of NIPT. The working group in charge of NIPT started from the collection of evidence and opinions. Between April and December 2016 it met with various stakeholders, including health professionals, organisations representing people with disabilities, as well as regulatory and

1775 ibid, p. 150. See also Brownsword and Wale, 'Testing Times Ahead' (2018) 81(4) Mod Law Rev p. 646, 668.

1776 Montgomery in Palazzani, *Role and Functions of Bioethics Committees* (2014).

1777 Hagedorn, *Legitime Strategien der Dissensbewältigung in demokratischen Staaten* (2013) p. 327; Montgomery in Palazzani, *Role and Functions of Bioethics Committees* (2014); Brownsword and Wale, 'Testing Times Ahead' (2018) 81(4) Mod Law Rev p. 646, 668

1778 Montgomery in Palazzani, *Role and Functions of Bioethics Committees* (2014). See also Brownsword and Wale, 'Testing Times Ahead' (2018) 81(4) Mod Law Rev p. 646, 669.

1779 Montgomery in Palazzani, *Role and Functions of Bioethics Committees* (2014); Brownsword and Wale, 'Testing Times Ahead' (2018) 81(4) Mod Law Rev p. 646, 649.

1780 "[T]he approach of the Nuffield Council on Bioethics […] has avoided a principle-based approach in favour of a procedural sense of legitimacy based on the inclusiveness of its listening processes, and the rigorous quality of the tests of rationality it applies to the arguments", Montgomery, 'Bioethics after Brexit' (2018) 18(2-3) Med Law Int p. 135, 153.

governmental bodies.[1781] An anonymous online survey was launched. In particular this sought to gather the opinions of individuals with personal and professional experience with NIPT. More than 700 people responded to the survey.[1782] In addition, the Council conducted an open public consultation between May and August 2016, asking for open answers to twenty questions. After spreading the consultation through social media and mailing lists, the Council received 28 responses from religious organisations, associations of people with Down's syndrome, medical societies, universities and others.[1783]

The outcome of the working group's activities was published in March 2017 in the report 'Non-invasive Prenatal Testing: Ethical Issues'. The document outlined an ethical framework based on the values of autonomy and consent, avoidance of harm, equality and inclusion.[1784] Having established the necessity to respect these principles, the Council clearly positioned itself in favour of the option endorsed by the UK National Screening Committee.[1785] The decision to offer NIPT to all women at high risk after initial screening was recognised by the Council as "a proportionate and ethical approach at the current time".[1786] This represents a compromise solution between protection of the woman's reproductive autonomy, avoidance of

1781 In June and July 2016, the Working Group met with healthcare professionals involved in delivering NIPT, charities representing people with genetic conditions and people with family members with genetic conditions, government, regulatory and professional bodies. Interviews were carried on with scientists working in areas relevant to NIPT, manufacturers of NIPT, women who had recently undergone NIPT, and people with genetic conditions; see Nuffield Council on Bioethics, 'Non-invasive Prenatal Testing: Ethical Issues', London 2017, pp. 145–147.

1782 ibid, p. 141.

1783 ibid, p. 143. The responses can be read in the document Nuffield Council on Bioethics, 'Non-invasive prenatal testing. Summary of consultation responses' (June 2017) <https://www.nuffieldbioethics.org/assets/pdfs/Analysis-of-NIPT-consultation-responses.pdf> accessed 24.3.2022.

1784 Nuffield Council on Bioethics, 'Non-invasive Prenatal Testing: Ethical Issues', London 2017. On the ethical starting points, see Brownsword and Wale, 'Testing Times Ahead' (2018) 81(4) Mod Law Rev p. 646, 649.

1785 „The Working Group supports the introduction of NIPT for Down's, Edwards' and Patau's syndromes in the NHS for women who have been found to have at least a 1 in 150 chance of having a fetus with one of these conditions", Nuffield Council on Bioethics, 'Non-invasive Prenatal Testing: Ethical Issues', London 2017, p. 134. See also Brownsword and Wale, 'Testing Times Ahead' (2018) 81(4) Mod Law Rev p. 646, p. 656.

1786 Nuffield Council on Bioethics, 'Non-invasive Prenatal Testing: Ethical Issues', London 2017, p. 134.

harm, and inclusiveness.[1787] On the one hand, offering the test guarantees the patient's right to information pertaining to her reproductive health. The Council acknowledged that the diagnosis enables the woman to make informed choices during pregnancy or, alternatively, to psychologically prepare and make practical arrangements for the birth of a child with a chromosome anomaly.[1788] In addition, the supply of NIPT is in line with the state's commitment to provide high-quality and safe health care.[1789] On the other hand, the highest accuracy of results must be ensured and NIPT must not be misused or used to diagnose non-significant or non-medical conditions.[1790]

The Council placed particular emphasis on the need to ensure women's informed consent, also mentioning the UK Supreme Court decision in *Montgomery v Lanarkshire Health Board*. High-quality information and support, including the communication that screening remains entirely voluntary, is considered essential for the ethical implementation of NIPT in the NHS.[1791]

Furthermore, the NCOB was concerned to ensure that the availability of the test did not lead to worse conditions for people with Down syndrome. The report pointed out that the impact of supporting children with disabilities on state resources cannot be included in the calculation of the cost-effectiveness of the test.[1792] In sum, the state must ensure that the

1787 Brownsword and Wale, 'Testing Times Ahead' (2018) 81(4) Mod Law Rev p. 646, 653.

1788 "The anxiety and uncertainty generated by a postnatal diagnosis relating to a lack of understanding about the condition and its implications, compounded by the physical aspects of childbirth and potential health threats to the baby, can make the assimilation of new information at this time extremely challenging. A prenatal diagnosis, on the other hand, can mean having time to understand and accept the diagnosis, to seek information and advice from support groups and other parents and to put any practical arrangements in place for after the birth, such as sourcing any special equipment or arranging additional childcare support. […] A prenatal diagnosis also allows medical interventions to be offered that can potentially improve the outcomes for the baby", Nuffield Council on Bioethics, 'Non-invasive Prenatal Testing: Ethical Issues', London 2017, pp. 52-53.

1789 ibid, p. 30.

1790 The prohibition of the use of NIPT to diagnose non-medical or less significant conditions represents an approach that some scholars have called paternalistic, albeit justifiable, see Brownsword and Wale, 'The Right to Know and the Right Not to Know Revisited' (2017) 9(1) Asian Bioeth Rev p. 3, 15.

1791 Nuffield Council on Bioethics, 'Non-invasive Prenatal Testing: Ethical Issues', London 2017, p. 128.

1792 ibid, p. 70.

implementation of the test remains in line with social equality and with the public support of people with disabilities.[1793]

b Considerations of Ethical Aspects by the UK National Screening Committee

In June 2017 the Nuffield Council of Bioethics' report on the ethical issue of NIPT was presented to the UK National Screening Committee.[1794] While supporting the decision to offer NIPT to women with a greater than 1 in 150 chance of having an affected foetus, the NCOB accused the UK NSC to have fallen short in considering the ethical aspects related to NIPT. The Council accordingly recommended that the UK NSC more fully consider the psychological, ethical and social consequences of prenatal screening programmes that could possibly lead to termination of pregnancy.[1795] In particular, the Nuffield report argued that attention should be paid to the possibility of passing unintended offensive messages towards people with disabilities. More generally, it was suggested that the UK NSC should develop ethical criteria for assessing screening programmes where abortion is an option and strengthen their public engagement activities as well as the representation of ethics experts on the committee.[1796] Associations representing people with disabilities have also denounced how an 'ethical vacuum' around the evaluation of prenatal screening has been made particularly visible through the case of NIPT.[1797] Feedback from a previous consultation conducted in 2015 by the UK NSC had already found insufficiencies in this regard and had suggested that the committee could benefit from the inclusion of additional members with expertise on ethical issues.[1798]

1793 ibid, p. 120.

1794 UK National Screening Committee, 'Note of the meeting held on the 23 June 2017' <https://www.gov.uk/government/publications/uk-nsc-meeting-june-2017> accessed 23.3.2022.

1795 Nuffield Council on Bioethics, 'Non-invasive Prenatal Testing: Ethical Issues', London 2017, p. 136.

1796 ibid.

1797 Intervention by Dr Elizabeth Corcoran at the Conference Prenatal Testing, Disability, and the Ethical Society, 'Reflections Following Crowter', 4.3.2022.

1798 UK National Screening Committee, 'Review of the UK National Screening Committee (UK NSC): Recommendations' (June 2015), p. 13 <https://www.gov.uk/government/uploads/system/uploads/attachment_data/file/443953/20150602_-_Final_Recommendations.pdf> accessed 23.3.2022: "Responses to the consultation

The UK NSC Guidance Criteria for appraising the viability, effectiveness and appropriateness of a screening programme had been updated in October 2015. This already stated the need to collect evidence that each screening programme would be socially and ethically acceptable to health professionals and the public in all its phases.[1799] Moreover, the Committee has always applied principles of deliberative democracy, by conducting public consultations and remaining fundamentally open to reviewing any decision should new evidence or any other arguments emerge.[1800]

However, as a result of the NCOB's recommendations on NIPT, the UK NSC has further intensified its focus on the ethical issues of prenatal screenings.[1801] For this purpose it has relied on the temporary transfer of a member of the Nuffield Council of Bioethics[1802] and on the recruitment of another permanent member with ethical expertise.[1803] The two new members assisted other committee members in setting up a new ethics task group within the UK NSC that was chaired by law professor Roger Brownsword.[1804]

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are clear that the UK NSC would benefit from additional ethical expertise, in particular there is support for drawing expert advice from a reference group of experts. Responses vary on whether this should be a standing organisation, a more ad hoc group or referring to external established ethical groups. The review group [...] acknowledges that sometimes particular expertise or a more focused consideration may be required".

1799 UK National Screening Committee, 'Criteria for appraising the viability, effectiveness and appropriateness of a screening programme' (23.10.2015), para. 4.12. <https://www.gov.uk/government/publications/evidence-review-criteria-national-screening-programmes/criteria-for-appraising-the-viability-effectiveness-and-appropriateness-of-a-screening-programme> accessed 23.3.2022

1800 Brownsword, 'Regulating The Life Sciences, Pluralism And The Limits Of Deliberative Democracy' [2010](22) SAcLJ p. 801, 822.

1801 As gladly noted by the Nuffield Council of Bioethics in Nuffield Council on Bioethics, 'Non-invasive Prenatal Testing: Ethical Issues. Review of Activities Since Publication', November 2018, p. 15.

1802 Joynson, 'Embedding ethics at the UK National Screening Committee' (23.3.2021) <https://phescreening.blog.gov.uk/2021/03/23/embedding-ethics-at-the-uk-national-screening-committee/> accessed 23.3.2022.

1803 UK National Screening Committee, 'Note of the meeting held on the 29 June 2018' <https://www.gov.uk/government/publications/uk-nsc-meeting-june-2018> accessed 23.3.2022. See also Nuffield Council on Bioethics, 'Non-invasive Prenatal Testing: Ethical Issues. Review of Activities Since Publication', November 2018, p. 15.

1804 Who was already a permanent member of the Committee. See UK National Screening Committee, 'Screening in the UK: making effective recommendations: 1 April 2017 to 31 March 2018' Ref: PHE gateway number 2018283, pp. 4–5 <https:/

An opportunity for the ethics task group to engage with NIPT presented itself in 2018, as the UK NSC received a proposal to include NIPT 'reflex testing' for chromosomal trisomies as part of the NHS Fetal Anomaly Screening Programme.[1805] This would imply that the future mother's blood sample to be used for NIPT would be collected already upon collection of the sample for the preliminary combined screening. Only if the result of the combined screening would reveal a probability of having an affected foetus of 1 in 800 would the second sample actually be used for NIPT. Such a procedure would in practice both reduce the eligibility threshold for NIPT and eliminate the need to recall the woman for a further consultation appointment and second blood sample collection after the combined test.[1806]

The responsibility for assessing this proposal was given to the newly established task group on ethics.[1807] Reporting back in October 2018 the task group advised the committee not to recommend the 'reflex' strategy for NIPT.[1808] They argued that such an approach raises several broad concerns regarding, *inter alia*, its suitability to support reproductive autonomy and its benefits in terms of resources savings. Moreover, the lower threshold for access to the test would lead to an expansion of its uptake, the ethical acceptability of which is, according to the task group, uncertain.[1809] The committee endorsed the group's assessment and agreed to waiting for the results of the evaluative roll out before making any adjustments to the NIPT screening pathway.[1810]

More generally, the work of the ethics task group has continued in the form of consultations with the public and stakeholders. The views gathered

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/assets.publishing.service.gov.uk/government/uploads/system/uploads/attachment_data/file/733226/Screening_in_the_UK_making_effective_recommendations_2017_to_2018.pdf> accessed 23.3.2022 See also Nuffield Council on Bioethics, 'Non-invasive Prenatal Testing: Ethical Issues. Review of Activities Since Publication', November 2018, p. 15.

1805 UK National Screening Committee, 'Note of the meeting held on the 29 June 2018'.

1806 ibid, paras. 3.8–3.10 for a description of the proposed procedure.

1807 ibid.

1808 UK National Screening Committee, 'Note of the meeting held on the 31 October 2018' <https://www.gov.uk/government/publications/uk-nsc-meeting-october-2018> accessed 23.3.2022.

1809 ibid, para. 3.13: "[T]here is uncertainty on whether expansion of the use of NIPT which would be a consequence of the strategy as currently proposed is ethically acceptable".

1810 UK National Screening Committee, 'Note of the meeting held on the 8 November 2019' <https://www.gov.uk/government/publications/uk-nsc-meeting-november-2019> accessed 23.3.2022.

have been used to develop ideas on how to improve procedures to more effectively include ethical considerations in the UK NSC's assessments.[1811] Most recently the ethics group worked to design an ethical framework for the analysis of screening programmes that was adopted[1812] and published[1813] by the UK NSC in 2021. This ethical framework is composed of four principles. They are aimed at: improving health and wellbeing, treating people with respect, promoting equality and inclusion and using public resources fairly and proportionately.[1814] For the improvement of health and wellbeing, benefits should be measured in terms of the individual to whom the screening is offered and should always prevail over potential harms. However, after ascertaining the potential benefits to the individual, the harms to others and to society can be considered. The principle of respect is specified in two ways. First, it implies that individual patients should be able to make fully informed screening choices aligned with their personal values. Second, it is argued that screening programmes "should take into account the views of those affected".[1815] According to the principle of equality and inclusion "any potential wider consequences of screening for society in the initiation and implementation of screening, both in the short and long term, should be considered".[1816] The ethical framework also calls for access to screening to be equitable and inclusive and for public resources to be used equitably and cost-effectively.

1811 "The ethics group would soon be engaging with external stakeholders and members of the public in order to gather views and experiences which would provide options on where, how and what is needed to engage, manage and allow for ethical considerations to be better incorporated into the UK NSC's processes", UK National Screening Committee, 'Note of the meeting held on the 28 October 2020' , para. 5.2 <https://www.gov.uk/government/publications/uk-nsc-meeting-october-2020> accessed 23.3.2022.

1812 UK National Screening Committee, 'Minutes 25 June 2021' <https://www.gov.uk/government/publications/uk-nsc-meeting-june-2021/uk-nsc-minutes-june-2021-draft> accessed 23.3.2022.

1813 UK National Screening Committee, 'UK NSC ethical framework for screening' (10.8.2021) <https://www.gov.uk/government/publications/uk-nsc-ethical-framework-for-screening/uk-nsc-ethical-framework-for-screening> accessed 23.3.2022.

1814 ibid.

1815 ibid.

1816 ibid.

c Room for Ethical Considerations in the Evaluation of Screening Programmes

The work that the ethics group has devoted to the publication of the UK National Screening Committee's ethical framework is to be welcomed insofar as it promotes the transparency of decisions that may be taken in the case of ethically controversial screening programmes. However, any normative framework influencing decision-making in the delivery of health services in England must be assessed in relation to the previously outlined procedural principles and in relation to the notion of accountability for reasonableness.[1817]

It must be borne in mind that the UK NSC is tasked with making recommendations to bodies, such as the Secretary of State and NHS England, that are bound by a legal and procedural framework. For example, the National Health Service Act 2006 in section 1A requires the Secretary of State to secure "continuous improvement in the quality of services". In particular, the provision of the services must ensure the outcomes of: service effectiveness, service safety and quality of patient experience. An equivalent duty to improve the quality of services is imposed on NHS England under section 13E of the NHS Act 2006, together with a duty to promote innovation in the provision of health services[1818] and a duty to enable patient choice.[1819] Both NHS England and the Secretary of State are also under an obligation to, respectively, promote the NHS Constitution[1820] and to have regard to it in exercising their functions.[1821] The NHS Constitution requires, *inter alia*, that the most effective use be made of scarce NHS resources. NHS patients have rights, albeit only procedural ones, that correspond to these obligations. Decisions concerning the design of screening programmes, as well as their public provision and commissioning, must be made with due regard to these obligations and procedural rights. NHS patients therefore have a procedural right to expect the authorities to strive to achieve this quality improvement. In the case of NIPT scientific evidence shows that its public funding would serve to improve the effectiveness and the safety of screening, as well as the quality of the patient experience.

1817 See Chapter 1, sec. B.III.2.b.
1818 National Health Service Act 2006 sec. 13K.
1819 National Health Service Act 2006 sec. 13I.
1820 National Health Service Act 2006 sec. 13C.
1821 National Health Service Act 2006 sec. 1B.

Furthermore, it was illustrated in the Chapter 1 how the commissioning of health services in England is subject to a procedural framework of accountability for reasonableness. Hereby decisions on the public funding of health services should be made by taking into consideration only relevant factors for the decision and avoiding grounds on which reasonable people might disagree.[1822] Thus, it is questionable whether ethical considerations that are only endorsed by a certain section of society and not widely accepted could legitimately play a role within this framework. Decisions that are binding on society as a whole primarily need to be taken in a manner that is coherent with a legal framework that all reasonable subjects can agree on.

Admittedly, as the Nuffield Council of Bioethics also noticed in its report on the ethical issues of NIPT, full consensus on all aspects of prenatal screening programmes is virtually unachievable.[1823] What is needed in order to comply with the English normative framework is to bring together the various ethical perspectives present in society and to try to reach a lowest common moral denominator on which to base the rules of the public healthcare system. In this context it is necessary to keep the public well informed about ethical aspects of new health services so that the constituency can express their informed opinion and so that the procedural legitimacy of the choices made by public authorities can be upheld. According to the UK constitutional framework legal measures are acceptable insofar as they respect procedural principles and, *inter alia*, remain flexible for challenges and amendments advocated by societal groups with diverging ethical views. This goal is facilitated, in the absence of an official national ethics council, by the work of the Nuffield Council of Bioethics.

The task of the UK NSC, however, seems to be a rather different one; namely to advise health ministers and the NHS on their decisions regarding, respectively, the design and implementation of screening. In fulfilling these functions it appears essential that all bodies involved be committed primarily to ensuring compliance with a framework of accountability for reasonableness.

This implies, first, that the principles and duties imposed on public bodies in designing the provision of health services must be adhered to. Such duties include those of quality improvement and respect for the NHS Constitution, as well as the standards of reasonableness and relevancy.

1822 See Chapter 1, sec. B.III.2.b.

1823 Nuffield Council on Bioethics, 'Non-invasive Prenatal Testing: Ethical Issues', London 2017, pp. 69-70.

Although it might be unlikely that a court in judicial review would overrule an ethically charged decision of a public body, such standards nevertheless politically constrain public authorities.

Second, compliance with the more general legal framework must also be respected, for example compliance with the rules on abortion and on patients' informed consent. This legal environment amounts to fundamental value decisions that are procedurally legitimate and generally binding and thus dictate substantive conditions, which are 'the embodiment of a common moral position', for the acceptability of screening programmes.

As far as NIPT is concerned this point has been elucidated clearly by Jeffrey Wale who rightly argues that "the purposes or aims of any prenatal testing regime need to be consistent with, and correlate to, the wider regulatory/legal framework in which that regime operates".[1824] In this sense, screening through NIPT should not be offered publicly for "purposes that would be or are likely to be incompatible with any framework for lawful abortion".[1825] The legal system must maintain its coherence.[1826] Conversely it follows that where NIPT meets all the criteria of quality, safety and effective use of public resources, with which the NHS and the Secretary of State are obliged to comply, its public funding could not reasonably be refused[1827] on the grounds of, for example, the ethical undesirability of abortion.[1828] The common moral position is represented by the Abortion Act 1967 and this remains open to amendments according to possible shifts in society's views.[1829]

1824 Wale, 'Don't Forget the Legal Framework' (2016) 15(4) Med Law Int p. 203, 209.

1825 ibid, p. 214.

1826 See also Brownsword and Wale, 'Testing Times Ahead' (2018) 81(4) Mod Law Rev p. 646, 662.

1827 This point is not shared by J. Wale, who claims that "even if a State provides lawful options to terminate pregnancy, it does not follow that those options should be encouraged via prenatal testing or otherwise through unlimited public funding", Wale, 'Don't Forget the Legal Framework' (2016) 15(4) Med Law Int p. 203, p. 210. This thesis concurs with the author only to the extent that unlimited public funding is certainly not mandatory. However, the possible rejection of inclusion within publicly funded medical care must come from allocation reasons and not from ethical considerations external to the legal system.

1828 This is because the common moral position is embodied in the Abortion Act 1967, which remains, admittedly, open to being amended in the event of changing views in society.

1829 This was ultimately recognised also by the opponents of NIPT. After losing the battle against public funding of NIPT, advocates tried to challenge sec. 1(1)(d) of the Abortion Act 1967 in front of the High Court of Justice for its alleged

In the same way the autonomy of the patient and their ability to make individual ethical choices is a legal value that must be maintained. This has two implications. On the one hand, patient autonomy is a legal constraint which requires that the inclusion of NIPT in the screening pathway is done with due regard to women's informed consent. It must be avoided, for instance, that the test is offered routinely without patients actually understanding which new information the test will provide and its consequences. On the other hand, however, the value of autonomy seems to conflict with a position of ethical paternalism in which the public provision of NIPT is rejected for fear that it may be contrary to the morality of a part of society.[1830] The refusal to include NIPT in NHS services would establish an economic barrier to accessing a more effective and safer screening that would otherwise facilitate equal access for all patients to an informed choice about their pregnancy.[1831]

In sum, the consideration of ethical aspects in the evaluation of screening programmes by the UK NSC can only be undertaken with the understanding that consistency with the regulatory framework must be maintained. Substantive concerns about NIPT should be resolved not by appeals to morality but by compliance with an approach that aims at protecting the interests of the various parties involved through a compromise widely acceptable to society as a whole.

If a number of ethical concerns remain inadequately protected by the statutory framework, then the latter might become the target of campaigns to promote amendments. Ethical and religious concerns, on the other hand, cannot significantly affect the arrangements for public coverage of health technologies, as this has to follow procedural principles whereby patients

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incompatibility with several provisions of the HRA Act, see *Crowter & Others, R v Secretary of State for Health And Social Care* [2021] EWHC 2536 (Admin) (23 September 2021). The challenge was, however, unsuccessful.

1830 The same view is endorsed by Nicholas Wald – albeit without employing legal reasoning – according to whom "[i]t is arguable that ethical review by a public agency [...] in respect of a screening programme deemed to be worthwhile [...] replaces individual choice with institutional decision making in areas where individual choice should prevail. [...]. Provided that a screening programme is lawful and is also justified on scientific and medical grounds, the individual is sovereign in determining the ethical position", Wald, 'Are Screening Practice Ethics Committees Needed?' (2021) 28(4) J Med Screen p. 377.

1831 On this point see Bunnik and others, 'Should Pregnant Women Be Charged for Non-invasive Prenatal Screening?' (2020) 46(3) J Med Ethics p. 194, 196; Bunnik and others, 'Why NIPT Should Be Publicly Funded' (2020) 46(11) J Med Ethics p. 783, 784.

cannot be denied access to health services on the basis of unreasonable or irrelevant factors.

As demonstrated in the case study on preimplantation genetic diagnosis, the legal criteria featured in the statutory framework already tend to accommodate the constituency's ethical concerns. Regarding the method of 'reflex testing' in the use of NIPT for example, this would not be compatible with the legal framework. This is not so much because it is 'ethically wrong' but rather because it does not seem to successfully respect women's informed consent and because the gathered scientific evidence suggests that offering the test to women with a chance of having an affected foetus of 1 in 800 would make screening less accurate and reduce the quality of the offer.[1832] Certainly, however, the reconstruction of the different ethical aspects is useful for informing the population and keeping the public debate open, thereby helping to maintain the legitimacy of public decisions, as the Nuffield Council of Bioethics has suggested.[1833]

These findings are relevant to the assessment of the ethical framework adopted by the UK National Screening Committee. The decisions of this committee are particularly influential for the shaping of screening programmes by the Secretary of State and their provision by NHS England. This influence is not only political but also legal since, according to the NHS Constitution to which the Secretary of State and the NHS must have regard, the NHS "commits to provide screening programmes as recommended by the UK National Screening Committee".[1834] In this way the consideration of ethical aspects in the decisions of the UK NSC is likely to be directly transposed into the choices of the public authorities following its recommendations. It is therefore desirable for the ethical framework adopted by the UK NSC to maintain consistency with the legal framework and accountability for reasonableness.

Depending on how the framework published in 2021 will be implemented in practice, the only problematic points in this respect are the reference to harms to others and to society and the consideration of "any potential

1832 Concerns about the allocative efficiency of public resources can admittedly also be described as 'ethical', but these fall beyond the scope of this dissertation.

1833 Nuffield Council on Bioethics, 'Non-invasive Prenatal Testing: Ethical Issues', London 2017, p. 70.

1834 Department of Health and Social Care, 'The NHS Constitution for England', 1.1.2021.

wider consequences of screening for society".[1835] As these statements are broadly open to interpretation they have the potential to be entry points for ethical considerations that are not compatible with the currently existing normative framework and thus function as 'Trojan horses'[1836] for ethical or religious considerations in the law.[1837]

D. Comparative Analysis

I. NIPT in the Private Sector

NIPT entered the European market as an IVD device in 2012. Not in all the three countries, however, has its mere entry onto the market triggered a large-scale public debate. Germany is the country where the discussions have been most heated ever since the introduction of this screening technology.[1838] An opinion of the German Ethics Council was requested as early as one year after NIPT was launched on the market, at a time when the G-BA had not yet expressed a position on whether or not an evaluation procedure for introducing this new technology into the statutory health insurance could be initiated.[1839] Germany is also the only one of the investigated countries where it was even disputed whether the test could be legally marketed. Reservations in this regard resulted from a legal expert opinion requested by the Federal Government Commissioner for Matters relating to Persons with Disabilities. The opinion stated that NIPT would endanger the health and safety of the foetus as a third party and that therefore its marketing should be prohibited.[1840]

In Italy and England too the rapid spread of NIPT on the private market has generated some apprehension. In contrast to Germany, however, such concerns were raised by specialised technical and scientific bodies. In Italy, for example, concerns have been expressed by the Italian National

1835 UK National Screening Committee, 'UK NSC ethical framework for screening', 10.8.2021.

1836 Using the term introduced by Spranger, *Recht und Bioethik* (2010) p. 38. See Chapter 1, sec. B.I.1.

1837 See Chapter 2, sec.D.IV.2.

1838 See above in this Chapter, sec. A.

1839 Deutscher Ethikrat, 'The Future of Genetic Diagnosis' (2013).

1840 Gärditz, 'Gutachtliche Stellungnahme zur Zulässigkeit des Diagnostikprodukts "PraenaTest"', 2012.

Health Council, which is responsible, among other things, for consulting the Ministry of Health on new health technologies.[1841] Moreover, in both countries worries about the use NIPT on the private market have focused on the possibly of poor quality information and counselling being given to patients by private facilities and not on the negative consequences for screened foetuses.[1842]

In sum, for as long as NIPT has been on the private market concerns about its uptake in England and Italy did not come close to reaching the broad scope of public discussion observed in Germany.

II. Public Coverage of Traditional Prenatal Testing

Having an overview of the traditional prenatal testing methods that are already included in the public coverage of the three jurisdictions provides insights into their general attitude towards public funding of screening for chromosomal aneuploidies.

A certain reluctance towards prenatal testing as part of the offer of the public healthcare system can be observed in Germany. Unlike in Italy and England the so called first-trimester screening or combined test is not included in the benefit basket of the statutory health insurance. This non-invasive screening technique can be performed at the patient's request, but the cost must be borne out-of-pocket. In England, on the contrary, combined screening is offered to all women in the first trimester of pregnancy, independently of their risk group. This is in line with NICE's recommendation that all women should be offered screening for chromosomal trisomies in the first trimester.[1843] In Italy as well, prenatal screening through combined testing has been offered free of charge to all patients starting from 2017.

With regard to invasive diagnoses, i.e. amniocentesis and chorionic villus sampling, these are offered to all patients found to be at high risk after combined testing in all three countries. In Germany a woman who is a first-time mother and over 35 is automatically considered to be at high

1841 Consiglio Superiore di Sanità, Sez. I, 'Linee-Guida. Screening prenatale non invasivo basato sul DNA (Non Invasive Prenatal Testing – NIPT)', 05.2015.

1842 Nuffield Council on Bioethics, 'Non-invasive Prenatal Testing: Ethical Issues', London 2017.

1843 National Institute for Health and Care Excellence, 'Antenatal care', 19.8.2021

risk. Among the analysed countries it is only in Germany that the public healthcare system's offer of these invasive diagnoses is seen as controversial by some legal scholars. They argue, in particular, that the detection of disability is not part of the statutory purpose of the health insurance.[1844] In Italy the National Bioethics Committee made an assessment of prenatal screening procedures in 2012 that was positive overall and argued that the right to know the health status of the foetus is undisputed, provided that couples are accompanied by a 'non-directive' medical consultation.[1845]

The different legal cultures of the three countries with respect to traditional prenatal screening or diagnosis are arguably reflected in their reaction to the emergence of more innovative testing tools. Indeed, Germany's skepticism towards classic prenatal testing methods resulted in a more heated public debate about NIPT.

III. Autonomy and Informed Consent

Legal principles protecting a woman's right to information on the health condition of her foetus are found in all three jurisdictions. These stem mainly from the protection of a patient's right to health, physical integrity and self-determination.

In Germany the state has an obligation to protect the right to physical integrity which includes, in the case of the pregnant woman, possible factors arising from the pregnancy that may affect her health. In Italy the right to health and the right to self-determination in matters of health receive special constitutional protection through a traditional patient-centred approach. It is assumed that knowledge of the foetus' health status strongly influences the pregnant woman's overall state of health. In England the value of patient autonomy has been given special consideration not least thanks to the intervention of the Supreme Court in 2015 in the landmark case of *Montgomery v Lanarkshire Health Board*.[1846] The improvement in reproductive autonomy is thus considered the main aim of prenatal testing.

Since its purpose is also the safeguarding of the woman's self-determination, prenatal testing should, according to the approach of all the analysed

1844 Welti in Becker and Kingreen, *SGB V* (2020) p. 254; Gärditz, 'Gutachtliche Stellungnahme zur Zulässigkeit des Diagnostikprodukts "PraenaTest"', 2012.
1845 Comitato Nazionale per la Bioetica, 'Diagnosi prenatali', 18.7.1992, pp. 28–31.
1846 *Montgomery v Lanarkshire Health Board* [2015] UKSC 11 (11 March 2015).

countries, only be offered if accompanied by adequate information and counselling. Its uptake is conditional on the informed consent of the patient, who must have fully understood the consequences and scope of the test about to be performed. It is clear in all jurisdictions that the information and counselling offered by the doctor should be of a non-directive nature, i.e. it should not aim to influence the woman and make her lean towards one choice over another. In the UK, for instance, it is emphasised that the value-based choices of individual patients must be protected and that healthcare professionals should not try to impose their ethical convictions on the patients.[1847]

This element was particularly relevant in Italy, where effectively implementing these informed consent principles was considered necessary and sufficient to overcome potential ethical concerns raised by NIPT. In contrast, a certain reluctance towards NIPT stems from the German approach placing particular emphasis on a woman's right not to know. This is not seen as a merely negative dimension of the right to know, but is considered an autonomous aspect of the right to self-determination in matters of health.[1848] This right is protected by the Genetic Diagnosis Act according to which the woman must be actively informed of her right not to know.

IV. NIPT in the Public Healthcare System

1. Criteria for Access to NIPT

The case study shows that there are several possibilities for designing prenatal screening programmes involving NIPT in a public setting. NIPT could be offered to all pregnant women or only to those in a certain risk category. In the second case, risk could be defined either by biological criteria, such as age, or by a previous screening test such as the combined test.

In Germany and England the public coverage of NIPT is now provided nationwide according to access requirements respectively established by the G-BA or suggested to the Secretary of State by the UK National Screening Committee.

1847 *Montgomery v Lanarkshire Health Board* [2015] UKSC 11 (11 March 2015).
1848 See, in this Chapter, sec. A.II.1.b.

In Germany a solution was found that requires an individual risk assessment for the specific patient. A statistically increased risk of trisomy would not be sufficient to access the test. Reimbursement is only granted when it is necessary to allow that particular woman to confront the possible presence of a chromosomal trisomy in the foetus within the framework of medical support.[1849] In other words, in order to obtain reimbursement from public health insurance funds the doctor and the patient must agree that in the individual case the uncertainty about the condition of the foetus is a disproportionate burden. Alternatively, the test can be accessed after a positive result from previous screening.[1850] This can be considered an acceptable compromise in that, on the one hand, it prevents the routinisation of the test but, on the other hand, it guarantees access to all women who consider NIPT necessary for the protection of their health.

In a similar fashion, the English UK NSC recommended NIPT for contingent use, i.e. dependent on the results of the combined test. Under its recommendations NIPT for trisomy 21, 13 and 18 should only be offered to women who exceed the risk threshold of 1 in 150 after the first screening.[1851] Accordingly, NIPT is not offered as standard testing to all women.

In Italy the access criteria for NIPT currently depend on the Region where the patient is resident. NIPT is still undergoing assessment by the Commission for the Updating of the LEA for its inclusion in the national Essential Levels of Care. However, the National Health Council has already issued guidelines, to which this commission can be expected to refer, which suggest that NIPT be offered for trisomies 13, 18 and 21 as a contingent screening after the combined test.[1852] As for the individual Regions, there is still some variety. Emilia Romagna has decided to offer free NIPT to all

1849 Gemeinsamer Bundesausschuss (G-BA), 'Beschluss über eine Änderung der Mutterschafts-Richtlinien (Mu-RL): Aufnahme einer Versicherteninformation zur Durchführung der Nicht-invasiven Pränataldiagnostik zur Bestimmung des Risikos autosomaler Trisomien 13, 18 und 21 mittels eines molekulargenetischen Tests (NIPT-Trisomie 13,18,21) für die Anwendung bei Schwangerschaften mit besonderen Risiken', 19.8.2021.

1850 ibid.

1851 UK National Screening Committee, 'UK NSC non-invasive prenatal testing (NIPT) recommendation', 01.2016.

1852 Consiglio Superiore di Sanità, Sez. I, 'Linee-Guida. Screening prenatale non invasivo basato sul DNA (Non Invasive Prenatal Testing – NIPT)', 05.2015; Consiglio Superiore di Sanità, Sez. I, 'Screening del DNA fetale non invasivo (NIPT) in sanità pubblica', 9.3.2021.

pregnant women regardless of risk factors.[1853] The self-governing province of Bolzano decided to provide NIPT only to patients who are found to be at intermediate risk after the combined test.[1854] In Tuscany as well, the test is only available to pregnant women who have been found to be at risk between 1/301 and 1/1000 after the combined test.[1855] Moreover, a small patient co-payment is applied to most patients here. Puglia statutorily introduced reimbursement of NIPT to all pregnant women who are either over the age of forty or found at high or intermediate risk after combined testing.[1856]

2. Ethical Concerns to Public Funding of NIPT

a Public Debates

In both England and Germany, the announcement that NIPT was being considered for introduction into the public healthcare system reinvigorated the public debate. Concerns about the possible effects of the use of NIPT were expressed in leading national media. In Germany an article in the newspaper *Zeit* denounced the introduction of NIPT into the GKV as the first step towards a society without people with disabilities.[1857] Similarly, in England a BBC documentary called out a possible drastic decrease in the number of children born with Down's syndrome.[1858]

In both jurisdictions these considerations also came to the attention of Parliament. In England the 'Don't screen us out' campaign was supported by a parliamentary motion.[1859] In Germany a parliamentary orientation debate on the issue of NIPT reimbursement by statutory health insurance was conducted in April 2019. On this occasion some MPs expressed the view that the state should not actively support any methods of screening people

1853 Regione Emilia-Romagna (Giunta Regionale), Delibera no. 1894, 4.11.2019.
1854 Provincia Autonoma di Bolzano - Alto Adige (Giunta Provinciale), Deliberazione no. 1413, 18.12.2018.
1855 Regione Toscana (Giunta Regionale), Delibera no. 1371, 10.12.2018.
1856 Legge Regionale Puglia no. 31/2021, "Implementazione del Test prenatale non invasivo (NIPT)" 6.8.2021.
1857 Bahnsen, 'Pränataldiagnostik: Der Test' *Die Zeit.* 22.1.2015.
1858 Phillips and Richards, 'A World Without Down's Syndrome', First Broadcast 5.10.2016 BBC.
1859 UK Parliament, 'Early Day Motion 44', 19.5.2016.

with chromosomal trisomies.[1860] They also noted that women must be guaranteed a right not to know and that discriminating messages towards people with disability must be avoided.

Groups advocating the ethically problematic nature of NIPT in both countries sought to influence the process of evaluating this technology for inclusion in the public healthcare system. In Germany the body responsible for deciding on the inclusion of NIPT in the statutory health insurance received letters from MPs twice. In the first one it was urged simply to consider the ethical and social consequences of NIPT in its assessment.[1861] In the second, more directly, the G-BA was asked to consider suspending the evaluation procedure because of ethical concerns.[1862] This and solicitations from other advocacy groups forced the authority to issue several statements on its awareness of the ethical issues of NIPT and to promise that the German Ethics Council would be involved in the decision.[1863] Even the technical body responsible for HTA in Germany was criticised for refusing to address possible ethical problems and devolving the consideration of such issues to the G-BA.[1864]

In England the parliamentary motion supporting the Down's syndrome community asked the government to postpone the introduction of NIPT in the NHS antenatal screening programme to allow further consultations.[1865] Responses to the public consultations have demonstrated, however, that the introduction of NIPT into NHS care, albeit with due caution, is ultimately relatively uncontroversial in the country. This view was also expressed by those legal scholars who intervened in the debate.[1866] It was observed that, in general, those members of society who are most critical of NIPT are also more generally opposed to any screening for chromosomal aneuploidies

1860 Deutscher Bundestag, 'Plenarprotokoll 19/95', Berlin 11.4.2019. See above, in this Chapter, sec. A.II.2.c.

1861 Hüppe and others, 'TOP 8.2.1 der 91. Öffentlichen G-BA Sitzung am 18. August 2016', 17.8.2016.

1862 As reported in the answer by the chairman of the G-BA, Gemeinsamer Bundesausschuss (G-BA), 'Schreiben von Prof. Josef Hecken, unparteiischer Vorsitzender des G-BA, an Mitglieder des Deutschen Bundestages zur Nichtvertagung der Beschlussfassung zu NIPT', 19.9.2019.

1863 Gemeinsamer Bundesausschuss (G-BA), 'Pressemitteilung Nr. 02/2015', 22.1.2015.

1864 Institut für Qualität und Wirtschaftlichkeit im Gesundheitswesen, 'IQWiG-Berichte - Nr. 623', 30.4.2018.

1865 UK Parliament, 'Early Day Motion 44', 19.5.2016.

1866 Brownsword and Wale, 'The Development of Non-Invasive Prenatal Testing: Some Legal and Ethical Questions' [2016](24) JRE p. 31.

that could lead to abortion. They believe that the public funding of such screenings sends an undesirable message by creating feelings of stigmatisation and discrimination in people with disabilities. The consensus of the general population, by contrast, seems to be that NIPT represents a fairly uncontroversial and beneficial innovation insofar as it limits the risks associated with invasive diagnoses that are already practised. This admittedly remains true only under certain conditions. First, NIPT should only be used for medical conditions that are already detectable by other screening techniques, such as chromosomal trisomies, and it should not be extended to purely aesthetic or non-medical conditions of the foetus.[1867] Moreover, respect of women's autonomy must be fully guaranteed.[1868]

Similarly, in Italy it is considered that the use of NIPT would not expand the uptake of prenatal screening to an unacceptable extent if it is limited to the detection of chromosomal trisomies.[1869] Here, in contrast to Germany and England, there has been no large-scale public debate on NIPT. The topic has only been addressed by bodies responsible for consulting the Ministry of Health or updating the Essential Levels of Care, a foundation dealing with women's health, and a few legal scholars.[1870] In Regions where NIPT has become part of the Regional Health System's benefit basket or where its evaluation for this purpose is ongoing, decisions on NIPT have generally been taken unanimously or almost unanimously. Those contributing to the Italian debate have agreed that the new moral issues emerging with NIPT can be effectively addressed by an adequate implementation of informed consent and counselling procedures in clinical practice.[1871]

1867 Brownsword and Wale, 'The Right to Know and the Right Not to Know Revisited' (2017) 9(1) Asian Bioeth Rev p. 3, 15.

1868 See above, in this Chapter, sec. C.II.1.b.

1869 Consiglio Superiore di Sanità, Sez. I, 'Linee-Guida. Screening prenatale non invasivo basato sul DNA (Non Invasive Prenatal Testing – NIPT)', 05.2015.

1870 See, *inter alia*, Consiglio Superiore di Sanità, Sez. I, 'Linee-Guida. Screening prenatale non invasivo basato sul DNA (Non Invasive Prenatal Testing – NIPT)', 05.2015; Consiglio Superiore di Sanità, Sez. I, 'Screening del DNA fetale non invasivo (NIPT) in sanità pubblica', 9.3.2021; Rizzo, 'Il consenso informato come strumento per l'implementazione etica dei test genetici non invasivi per la diagnosi prenatale' [2018](3) BioLaw Journal – Rivista di BioDiritto p. 225; Palazzani, *Dalla bio-etica alla tecno-etica* (2017).

1871 Palazzani, *Dalla bio-etica alla tecno-etica* (2017); Rizzo, 'Il consenso informato come strumento per l'implementazione etica dei test genetici non invasivi per la diagnosi prenatale' [2018](3) BioLaw Journal – Rivista di BioDiritto p. 225;

This comparatively uncontroversial approach to public funding of NIPT can be explained, firstly, by a relatively positive attitude to traditional prenatal diagnoses. Compared to them, NIPT is regarded as merely an improvement in the interests of both the future mother and the foetus. This attitude is in line with the Italian constitutional approach to the right to health as a fundamental right of every individual which is connected to their most intimate sphere. The fact that a potential public funding of NIPT does not lead to ethical conflicts could be linked to the idea that the National Health Service aims at protecting the core of the right to health, combined with a traditionally very broad conception of the notion of health.[1872] It follows that all health services pertaining to this essential core, which include prenatal diagnoses because of their importance for the psycho-physical well-being of the mother, are worthy of being equally guaranteed to all residents. This conception of the benefits provided by the National Health Service differs from the traditional conception of the German healthcare system, which is seen as an insurance scheme covering specific health risks.

b Consideration of Ethical Concerns in the Evaluation Procedure

i. Procedural Aspects

Although to varying degrees, ethical considerations were accounted for in the process that led (or is leading) to the inclusion of NIPT in the coverage of the public healthcare system in all three jurisdictions.

With regard to procedural elements used in dealing with ethical concerns in the three jurisdictions, it is not surprising that the element of public and stakeholder consultations played a particularly essential role in England. The UK National Screening Committee, the body in charge of evaluating screening programmes, decided to launch a three-month public consultation before issuing its final recommendation on NIPT.[1873] Thirty stakeholders with very different backgrounds and perspectives responded

Consiglio Superiore di Sanità, Sez. I, 'Screening del DNA fetale non invasivo (NIPT) in sanità pubblica', 9.3.2021.

1872 On the broad definition of health endorsed by Italian Constitutional Law, see Introduction and Chapter 1, sec. B.II.2.

1873 UK National Screening Committee, 'Note of the meeting held on the 18 June 2015'.

to this consultation. In the aftermath of the approval of NIPT for inclusion in NHS care, and as a response to the call to strengthen the consideration of ethical aspects, an ethics task group was set up within the UK NSC and this also worked through consultations with the public and stakeholders. In addition, the government has twice been called upon to consult more appropriately with the community of people with Down's syndrome before providing for public reimbursement of NIPT.[1874] This demonstrates the importance of consultation as an essential element of the legitimacy and acceptability of decision-making in England.

Also the Nuffield Council of Bioethics, when dealing with the ethical issues of public funding of NIPT, first of all engaged in public consultations and collection of stakeholders' opinions. The Council also launched an anonymous online survey to reach the opinions of individuals who had dealt with the test through personal or work experience.[1875] When drafting its NIPT report the Council followed its traditional method of applying criteria of procedural legitimacy and standards of reasonableness.

Public consultations did not have the same relevance in Germany and Italy. However, the German G-BA did use its formal consultation procedure to get the opinion of various stakeholders. It also sought to widen the debate by asking for comments from the German Ethics Council and the Genetic Diagnostics Commission.[1876] Moreover, the authority encouraged Parliament to initiate a discussion on the political and normative aspects of NIPT and directly interacted with stakeholders and the general public through its press releases. The IQWiG also conducted a public consulta-

1874 UK Parliament, 'Early Day Motion 44', 19.5.2016; Ravitsky, 'The Shifting Landscape of Prenatal Testing' (2017) 47(Suppl 3) Hastings Cent Rep S34-S40, S37.

1875 Nuffield Council on Bioethics, 'Non-invasive Prenatal Testing: Ethical Issues', London 2017.

1876 Gemeinsamer Bundesausschuss (G-BA), 'Beschluss des Gemeinsamen Bundesausschusses über die Einleitung des Stellungnahmeverfahrens gemäß § 91 Absatz 5, § 91 Absatz 5a sowie § 92 Absatz 1b und § 92 Absatz 7d des Fünften Buches Sozialgesetzbuch (SGB V) vor einer abschließenden Entscheidung über eine Änderung der Mutterschafts-Richtlinien: Nicht-invasive Pränataldiagnostik (NIPD) autosomaler Trisomien 13, 18 und 21 mittels eines molekulargenetischen Tests (NIPT) für die Anwendung bei Risikoschwangerschaften im Rahmen der Mutterschafts-Richtlinien (Mu-RL)', 22.3.2019; Gemeinsamer Bundesausschuss (G-BA), 'Nicht-invasive Tests bei Risikoschwangerschaften: G-BA fordert zur Stellungnahme auf', 22.3.2019.

tion open to all interested individuals, institutions and organisations.[1877] Furthermore, in February 2022 – on the occasion of the approaching implementation of NIPT reimbursement by health insurance funds – the German Ethics Council revived the debate by holding an online public discussion on NIPT with public participation via online questions.[1878] It can thus be observed how elements of the procedural model were incorporated into the decision-making procedure for NIPT in Germany and contributed to the achievement of an acceptable compromise regarding its reimbursement scheme.

ii. Substantive Elements

In terms of substantive considerations, in all three countries the bodies responsible for evaluating NIPT for public funding focused primarily on ensuring that women are not pressured into taking the test or into making any particular choice after a positive result. Reproductive autonomy and informed consent were the main theme throughout this case study and guaranteed acceptability of public funding for NIPT. Partially related to this, the need to avoid routinisation of NIPT was also addressed.

The element of reproductive autonomy and informed choice was of decisive significance especially in Italy. As demonstrated above,[1879] the Italian debate indicated that respect for the woman's informed consent, accompanied by adequate counselling, could be a necessary and sufficient condition to overcome any doubts about the desirability of publicly funding NIPT. Along the same lines the National Health Council also argued that the way to settle the ethical concerns raised by NIPT would be through non-directive counselling, offered as part of the diagnostic treatment.[1880] The focus of public decision-makers has thus been on maximising respect for women's reproductive autonomy. The overarching consensus on informed consent has here prevented the emergence of ethical controversies over NIPT.

1877 Institut für Qualität und Wirtschaftlichkeit im Gesundheitswesen, 'Nicht invasive Pränataldiagnostik (NIPD) zur Bestimmung des Risikos autosomaler Trisomien 13, 18 und 21 bei Risikoschwangerschaften (Vorbericht)', 11.12.2017, p. III.

1878 Deutscher Ethikrat, 'Pressemitteilung 01/2022'.

1879 See above, in this Chapter, at sec. B.3.

1880 Consiglio Superiore di Sanità, Sez. I, 'Screening del DNA fetale non invasivo (NIPT) in sanità pubblica', 9.3.2021, p. 21.

In England too the decision-making process has taken into account the need to avoid misleading information and the application of inappropriate pressure to patients. The RAPID study, initiated with input from the UK NSC, aimed not only at considering the scientific accuracy of the test, but also to assess the possibility of maintaining high levels of informed choice.[1881] NIPT was then recommended for introduction into NHS care on an evaluative rather than a permanent basis in order to better understand the impact of public funding on patients' reproductive autonomy. The UK NSC then used the argument of reproductive autonomy to reject the proposal received in 2018 to include the 'reflex testing' method for NIPT in the prenatal care pathway. It is to be noted, however, that the uncertain ethical assessment of a possible expansion in the uptake of NIPT, given the lower risk threshold for access, also played a role in this appraisal.[1882] Here too, the inclusion of an evaluation period and the emphasis on informed consent ensured the acceptability of the final compromise.

In Germany, the G-BA effectively addressed concerns about the right to know and not to know. The authority confirmed that NIPT can only be performed after giving the patient comprehensive counselling and information as well as sufficient time for reflection.[1883] Moreover, the possible routinisation of NIPT was prevented when setting the requirements for access to the test. Statutory health insurance coverage is provided only after an individual assessment of the woman's situation and her personal need to obtain information on the health status of the foetus.[1884]

As regards the other ethical concerns raised, they were not explicitly targeted by the relevant decision-making authorities. The G-BA in Germany emphasised that they had followed the legally prescribed procedure of evaluating the test and stressed that any remaining ethical issues at stake would require a legislative response.[1885] In England the issue of a possible increase in abortion rates was tackled by the RAPID study. With regard to the negative signals allegedly sent to the disabled community by the provision of public funding for NIPT, the UK NSC responded by including certain

1881 Hill and others, 'Evaluation of Non-invasive Prenatal Testing (NIPT) for Aneuploidy in an NHS Setting' (2014) 14(229) BMC Pregnancy Childbirth p. 1.
1882 UK National Screening Committee, 'Note of the meeting held on the 31 October 2018'.
1883 Gemeinsamer Bundesausschuss (G-BA), 'Beschluss über eine Änderung der Mutterschafts-Richtlinien (Mu-RL)', 19.9.2019 BAnz AT 20.12.2019 B6.
1884 ibid.
1885 Gemeinsamer Bundesausschuss (G-BA), 'Pressemitteilung Nr. 26/2019', 19.9.2019.

principles in its ethical framework for the assessment of new screening methods. These include: the possibility to consider harms to others and to society, as well as any potential wider consequences of the implementation of the screening for society.[1886]

c Assessment

i. Compliance with the Normative Framework

When considering the high accuracy, safety and cost-effectiveness of NIPT it seems that any refusal to include it in the public healthcare system could only be based on ethical or religious considerations. From a legal point of view these non-invasive technologies are no different from other existing prenatal screening methods. They simply better protect the rights of the foetus and increase the quality and safety of health services. The mere possibility of the test being used more often does not create any discrimination against people with disabilities. Discrimination would be caused, if anything, by the woman's subsequent choice to have an abortion. This choice, however, can legitimately be made if it remains within the statutory agreement and constitutional balance reached in each jurisdiction. The argument that an increase in the use of prenatal diagnostics would be undesirable is based on purely ethical and not legal grounds.

Thus, for each country an assessment can be made to determine whether purely ethical concerns could legitimately result in a decision not to publicly fund NIPT.

First, it results from the considerations made within the case study, that in all three jurisdictions the evaluation of NIPT for inclusion in the public healthcare system should primarily be carried out in accordance with the legal framework.[1887] In Italy and Germany substantive legal principles that govern the updating of the health benefit basket remove any room for the consideration of purely ethical concerns in the decision-making process.

In Germany the decision must be made on the basis of the aspects defined in § 135(1) of the SGB V. Namely, diagnostic or therapeutic benefit, medical necessity and economic efficiency. The list of criteria contained in

1886 UK National Screening Committee, 'UK NSC ethical framework for screening', 10.8.2021.

1887 See in this Chapter, for Germany sec. A.II.3, for Italy sec. B.II.3.c, and for England sec. C.II.3.c.

this paragraph is exhaustive and there is no legal basis that would allow the G-BA to bring ethical aspects into consideration. Neither could the G-BA legitimately block the procedure or postpone the decision on ethical grounds.

In Italy, a health service falls under the Essential Levels of Care when it is necessary to uphold the 'inviolable' core of the fundamental right to health throughout the national territory. Inclusion of a new health technology in the benefit basket follows the criteria set out in Article 1(2) of Legislative Decree no. 502 of 30 December 1992, and in particular those of quality of care, appropriateness and economical use of resources.

In England too there is rather limited space for the influence of purely ethical concerns, although this stems mainly from the particular pragmatic and procedural approach surrounding the public funding of new technologies in the NHS. English health authorities tend to take pragmatic decisions and to comply with a procedural model of 'accountability for reasonableness'. Moreover, the Secretary of State and NHS England are bound to respect procedural rights of the patients, as enshrined in the National Health Service Act 2006 and the NHS Constitution. These require, *inter alia*, that the state continuously pursues the improvement of quality of health care.

Beyond the legal criteria specifically drafted for the updating of the services provided by each healthcare system, each analysed jurisdiction is embedded into an overarching constitutional framework which would still preclude purely ethical considerations from negatively influencing the public funding decision for new health technologies.[1888]

In the case of NIPT, all three jurisdictions have proven their commitment to their legal and constitutional frameworks in deciding on the public coverage of NIPT.

In Germany, there has been no violation of the principle of ethical neutrality of the state in the G-BA's decision to introduce NIPT into the maternity guidelines. The instruments used to ensure the acceptability of the decision in this ethically controversial issue were mainly substantive and legal, although procedural mechanisms and dialogue with the public were implemented by the G-BA and the German Ethics Council. In explaining the reasons for its decision the G-BA focused mainly on the legal criteria regulating the inclusion of new products in the benefit basket of the GKV according to § 135(1) no. 1 SGB V. The provision of appropriate

1888 ibid.

counselling and information is a legal requirement that derives its validity from constitutional norms on self-determination and bodily integrity. This is reflected in the rights to know and not to know. Also the decision to only grant reimbursement of NIPT after individual assessment of each patient's case can be considered justified on the basis of these two constitutional interests. In sum, ethical considerations did not ultimately influence the reimbursement decision, which was lawfully made by the G-BA following the procedure set out in § 135 of the SGB V. The only factor that might have been negatively affected by the ethical weight of this topic is the timing of the decision. Given the demands from society and from members of Parliament the G-BA felt compelled to leave enough time for the legislature to intervene independently on the matter.[1889] This might have resulted in a lengthening of the timeframe needed for the decision.[1890]

The principle of laicity has been respected in the Italian case. The regional and national public authorities involved to date have upheld the constitutional principle of informed consent, as enshrined in the Constitution under the combination of Articles 2, 13 and 32. Any ethical issues relating to NIPT were held to be resolvable by protecting patients' ability to give informed consent, thus demonstrating a laicity-driven approach to the balancing of interests between the woman and the foetus. The final decision of the Commission for the updating of the Essential Levels of Care has not yet been reached, but it can be expected that it will rely on the guidelines of the National Health Council. These have stressed that the inclusion of NIPT in the Essential Levels of Care is necessary to ensure compliance with the criterion of appropriateness and to prevent carrying out riskier diagnoses.[1891]

In sum, both Italy and Germany based the neutrality and legitimacy of their decisions on NIPT primarily on substantial legal considerations.

1889 As sustained by the chairman of the G-BA in his letter to the MPs, the G-BA "has initiated a formal method evaluation procedure and conducted an extended (public) comment procedure in order to create time and space for parliamentary decision-making and, if necessary, also a parliamentary decision", see Gemeinsamer Bundesausschuss (G-BA), 'Schreiben von Prof. Josef Hecken, unparteiischer Vorsitzender des G-BA, an Mitglieder des Deutschen Bundestages zur Nichtvertagung der Beschlussfassung zu NIPT', 19.9.2019.

1890 While the procedure was first initiated in 2014, the final decision arrived in 2019 and the reimbursement from the health insurance funds will only be granted starting from spring 2022.

1891 Consiglio Superiore di Sanità, Sez. I, 'Screening del DNA fetale non invasivo (NIPT) in sanità pubblica', 9.3.2021.

Unsurprisingly, mainly procedural means were used in England to ensure that the decision remained legitimate and widely acceptable despite dealing with a highly ethically controversial question. This is consistent with the principles of procedural legitimacy underlying the English constitutional system. For example, the public was able to participate in the debate on the ethical aspects of the inclusion of NIPT in NHS care through consultation exercises from the UK NSC and the Nuffield Council of Bioethics. The opinions thus collected were put to a test of reasonableness. As far as the substantive considerations weighing on the decision are concerned, they were in line with the principles of accountability for reasonableness and other statutory requirements. All factors that weighed on the final decision could be broadly regarded as relevant and reasonable. Indeed, reasonable criteria of quality and accuracy were used and reference to factors on which reasonable people might disagree was avoided. The adherence to legal criteria ensured that the decision was in line with factors widely accepted as relevant, such as the "continuous improvement in the quality of services" established by the National Health Service Act 2006.[1892] Inclusion of NIPT in NHS antenatal care for patients at high risk after combined testing, in order to avoid invasive and harmful diagnoses, upholds the procedural rights of patients to the improvement of both the effectiveness and the safety of screening. Patients' procedural rights under the NHS constitution were also respected. This requires the government to provide screening programmes as recommended by the UK National Screening Committee. The government fulfilled this obligation by refusing to further delay the implementation of NIPT in the NHS.[1893]

ii. Calls for More Consideration of Ethics in the Decision-Making

In England and Germany the public funding of NIPT has triggered calls for a better inclusion of ethics in the assessment procedure leading to the public coverage of health technologies.[1894] However, if the normative framework of neutrality of justification is to be maintained, then it is not

1892 National Health Service Act 2006 sec. 1A.

1893 Lavery, 'Pregnancy: Screening. Question for Department of Health and Social Care', 2.9.2019.

1894 See criticism of G-BA and IQWiG in Germany (in this Chapter, sec. A.II.2.b) as well as the proposals for better inclusions of ethics in the UK NSC assessment of screening programmes (in this Chapter, sec. C.II.3.b).

possible to authorise public decision-makers to refuse the reimbursement of health technologies on purely ethical grounds. This consideration is valid for all three examined jurisdictions.

The German G-BA could only have blocked or delayed the evaluation procedure on the grounds that the increased use of prenatal diagnosis was concerning if the legislature had given it the competence to assess ethical or religious aspects. However, the suggestion that the legislature in future might allow the G-BA to consider ethical aspects in its evaluation procedure[1895] creates fertile ground for an infringement of the principle of neutrality of justification. Granting competence in ethical matters to the self-administration authority of the statutory health insurance could serve as a 'Trojan horse' for considerations linked to one specific ethical or religious belief in reimbursement decisions. Such justifications are not acceptable under a constitutional framework where the principle of ethical and religious neutrality also applies to the choices made by the welfare state in its action to implement the public healthcare system. In sum, any justification for refusing public funding purely based on ethical or religious views must be considered purely arbitrary and as contrary to the principle of ethical neutrality of justification.

In England too the ethical framework recently adopted by the UK NSC has a potential to function as 'Trojan horse' for ethical or religious considerations in the law.[1896] This would happen if one or more of the principles of this framework were interpreted according to a perspective that was not widely shared and if this were used to impose a particular belief without it being subject to the procedural principles that determine its legitimacy and acceptability by society as a whole. This could also lead to irrelevant factors or unreasonable considerations being taken into account in the decision-making of this public body. A consideration of non-widely shared ethical principles in the decisions of health authorities would thus run against the procedural principles of English constitutional law and the requirements of the model of accountability for reasonableness. Admittedly, however, the pragmatic and utilitarian positioning of the UK NSC seems to exclude this possibility, at least for the time being.

1895 Huster, 'Non-invasive Prenatal Diagnostics (NIPD) in the System of Medical Care' (2021) 49(8) J Perinat Med p. 1; Gemeinsamer Bundesausschuss (G-BA), 'Schreiben von Prof. Josef Hecken, unparteiischer Vorsitzender des G-BA, an Mitglieder des Deutschen Bundestages zur Nichtvertagung der Beschlussfassung zu NIPT', 19.9.2019.

1896 See, in this Chapter, sec. C.II.3.c.

Such calls for a greater consideration of ethical aspects in the health technology assessment process did not occur in Italy in the case of NIPT, given the relatively uncontroversial nature of this new prenatal screening. Also in this jurisdiction, however, a consideration of the 'ethical desirability' of new technologies could only be legitimately influential on decision-making if this ensured the compliance of new health services with widely accepted fundamental principles laid down in legislative acts that were themselves in line with the Constitution. This was the definition of ethical desirability endorsed by the previous Commission for the updating of the LEA.[1897] This strictly secular definition of 'ethical concerns' would be compatible with the Italian normative and constitutional framework of laicity.

1897 Arcà and Cislaghi, 'Percorsi metodologici per l'inserimento o l'esclusione di una prestazione dai Livelli essenziali di assistenza' [2006](2) Tendenze nuove p. 97, 102; Commissione nazionale per la definizione e l'aggiornamento dei Livelli essenziali di assistenza in Falcitelli and Langiano, *La remunerazione delle attività sanitarie* (2007) p. 254.