Hellenic Republic
National Commission
for Bioethics & Technoethics

RECOMMENDATION

On the bioethical dimensions of newborn genetic screening by human genome sequencing

November 2025

NATIONAL COMMISSION FOR BIOETHICS & TECHOETHICS
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I. Introduction

It is known that since 1974 Greece has been implementing the National Newborn Screening Program (N.N.S.P.)[1] with the aim to facilitate early diagnosis of rare, severe, manageable or treatable (e.g., congenital hypothyroidism, phenylketonuria, galactosaemia, G6PD deficiency). The N.N.S.P. is carried out by the Institute of Child Health (I.C.H.)[2], it encompasses all newborns in the country and is offered free of charge. The screening involves analyzing specific substances and biomarkers in a few drops of blood collected from a heel prick (via scarification) on the newborn's 3rd-4th day of life), enabling the detection of congenital metabolic disorders. These disorders typically do not present with observable phenotypic manifestations during the first days of life. If not detected in time, they can lead to severe metabolic issues, and/or intellectual disability, cerebral palsy, or even sudden death. Early diagnosis (within one month of life) and timely treatment, often through simple dietary adjustments or medication, can prevent disease manifestation and ensure a good quality of life. The N.N.S.P. is not limited to measuring a chemical substance but also provides a comprehensive service that includes detection confirmation, diagnosis, treatment, and monitoring of the child's progress, in line with the criteria of the World Health Organization (WHO).

The development of new detection methods, including biochemical and molecular approaches, and the rapid advances in medical genetics and genomics, have enabled the expansion of newborn screening. As a result, screening for hereditary disorders is evolving towards comprehensive genomic screening. Ongoing technological advances and declining costs of whole-genome sequencing make it technically feasible to identify the current 6,630 known monogenic (rare) diseases caused by variants in more than 4,670 genes[3], hundreds of which are now manageable or even treatable due to advances in genetics, biochemistry, gene therapy, and targeted genetic/genomic therapies[4].

II. Benefits

By analyzing the genome of newborns, it is possible to identify the potential for sever monogenic (rare) diseases, thereby serving both the individual's right to health, particularly in terms of disease prevention and protection from health harm, and the social right to health, as it enables the state to implement timely measures to

[1] https://www.eppen.gr
[2] https://ich.gr/
[3] https://www.omim.org/statistics/geneMap
[4] https://pmc.ncbi.nlm.nih.gov/articles/PMC11331404

prevent such diseases or, at a minimum, to treat them more effectively should they occur.

In parallel, the goal of a genomic screening program is to determine the prevalence of monogenic diseases within the population. In Greece, particularly due to migration patterns and potential cases of consanguinity, genetic variants previously undetectable in the population, and the associated hereditary diseases, may now emerge.

The substantial benefits for newborns, their families, and public health arise under clear guarantees of scientific reliability and subject to ethical, professional, deontological, and legal prerequisites. In the international literature, codes of ethics and guidelines have been developed and are continuously updated, which should underpin approved genetic screening programs[5]. In the present Recommendation, the Hellenic National Commission for Bioethics and Technoethics sets out the ethical, legal, and functional principles that should govern newborn genetic screening.

To ensure the most comprehensive possible briefing on the parameters that need to be taken into account with regard to the subject of the Recommendation, the Commission sought the opinions of scientists of recognized standing and representatives of relevant bodies through the submission of pertinent memoranda (see Annex).

III. Ethical principles

According to the Commission, the regulatory pillars on which every newborn genetic screening program must be based are the following:

a) **The principle of autonomy**, which is linked to **human dignity**, requires that parents' ability to decide freely whether their child will undergo genetic screening be safeguarded, following their free and thorough information about the conditions under which such genetic screening is conducted and about the protection of the newborn's sensitive personal data. This is because the collection of large volumes of genetic data objectively increases the risk of potential future misuse, even where appropriate protective measures have been taken. Explicit

[5] See, for example, Wilson, J. M. G. & Jungner, G. (1968). *Principles and Practice of Screening for Disease*. Geneva: World Health Organization, Public Health Papers No. 34. Kelleher KJ, et al.. *Principles for Primary Care Screening in the Context of Population Health*. Acad Pediatr. 2024 Jul;24(5):719-727. doi: 10.1016/j.acap.2024.02.015. Brlek P, Bulić L, Bračić M, Projić P, Škaro V, Shah N, Shah P, Primorac D. *Implementing Whole Genome Sequencing (WGS) in Clinical Practice: Advantages, Challenges, and Future Perspectives*. Cells. 2024 Mar 13;13(6):504. doi: 10.3390/cells13060504. PMID: 38534348; PMCID: PMC10969765. Basel-Salmon L, Brabbing-Goldstein D. *Fetal whole genome sequencing as a clinical diagnostic tool: Advantages, limitations and pitfalls*. Best Pract Res Clin Obstet Gynaecol. 2024 Dec;97:102549. doi: 10.1016/j.bpobgyn.2024.102549. Epub 2024 Sep 3. PMID: 39259994.

consent, following specific information (opt-in), must, as a matter of principle, precede the conduct of any type of testing (biochemical and/or genetic) and the collection of the newborn's data. Moreover, over time, increasing weight should be given to the views of the child itself regarding the retention and future use of its genetic data, and upon reaching adulthood, the individual should be able to revoke the parents' consent concerning the retention or any use of such data.

Furthermore, taking into account that the majority of high-throughput genetic analyses are carried out by a small number of specialized companies abroad, the Commission does not exclude a prospective collaboration with private laboratories that possess appropriate equipment – a practice that is well-established internationally and is already applied in Greece. However, in such cases, the principle of human dignity additionally dictates the following: contracts between the Ministry of Health or Legal Persons of Public Law and private laboratories operating in the field of genetic testing cannot have as their object the "ownership" of personal data, due to their connection to the personality of the data subject. Consequently, no agreement may provide for the transfer the "ownership" of personal data to a private laboratory in exchange for the services it provides. The private laboratory is remunerated by the contracting authority (the Ministry or another public body) solely for the services it provides, without thereby acquiring proprietary rights over the data themselves (or over the biological material from which they derive). The contracting authority holds no property rights over the data either.

b) The principle of beneficence, according to which every newborn genetic screening program must have demonstrated clinical utility, which presupposes the existence of sufficient scientific evidence justifying the choice of a treatment or of preventing measures. The collection of genetic information is ethically justified only when it can be proven to be beneficial for the protection of the future health of the children undergoing genetic screening.

c) The principle of non-maleficence. In all cases, a genetic screening program must entail any risk of causing harm to physical or mental health, including the future health of the screened newborn. Potential risks of the "over-medicalization" of children's lives as a whole, resulting from their inclusion in preventive screenings for possible future risks of rare diseases, must be taken into account and addressed through appropriate measures, including the provision of genetic counselling. At the same time, emphasis must also be placed on addressing the potential psychological impacts that such screenings may have on parents and families of newborns, etc.

d) The principle of fairness requires that public health policies involving newborn genetic screening be formulated on the basis of fair distribution, equal access for all concerned, and effective protection against unethical uses that could stigmatize individuals or lead to negative discrimination or social exclusion. Whole-genome analysis of newborns generates data not only about the newborn, but also

about their family that may potentially be used in the future by third parties for purposes beyond those of the initial examination, and indeed without the consent of the individual concerned or the parents. It is essential that all measures be taken to ensure that those involved do not suffer discrimination in employment or in life and health insurance on the basis of their genetic predispositions.

IV. European Legal Reference Framework

According to the Commission, any genetic screening program, whether applied to the whole population or to a section of population, should comply with the conditions set out in *the Additional Protocol to the Oviedo Convention on Genetic Testing for Health Purposes* (27.11.2008)[6]. The Commission notes that although the Protocol has not been signed and ratified by Greece and is therefore not legally binding, it constitutes a set of soft-law norms and an established bioethical acquis that cannot be disregarded.

More specifically, Article 19 of the Protocol, permits, in principle, the implementation of a genetic screening program for health purposes, provided that is approved by a competent body of the Member State. Such approval must be granted following an independent evaluation of the program's ethical acceptability and fulfilment of specific conditions, laid down in the Protocol. These conditions must be assessed individually, on a case-by-case basis[7]:

(a) Recognition of the health relevance of the program for the whole or section of the population concerned (in other words, the condition targeted by the genetic test should constitute a significant public health issue either because of its severity or because of the number of individuals affected).[8]

(b) Establishment of the scientific validity and effectiveness of the program; for example, a demonstrable reduction in overall mortality or morbidity constitutes a key criterion for the effectiveness of a population-based screening program.[9]

(c) Availability of appropriate preventive or treatment measures for the disease or disorder being screened. Prevention should be understood in a broad sense and may include, for instance, closer medical monitoring through more regular examinations, measures that limit or delay the progression of the symptoms of a disease and significantly improve the well-being or living conditions of the individual,

[6] Additional Protocol to the Convention on Human Rights and Biomedicine concerning Genetic Testing for Medical Purposes (CETS No. 203).
[7] Explanatory Report to the Additional Protocol, §145.
[8] Explanatory Report to the Additional Protocol, §147.
[9] Explanatory Report to the Additional Protocol, §148.

while a measure may even be a reproductive choice when a severe disease is difficult to treat.[10]

(d) Provision of appropriate measures to ensure equal access to the program (the concept of equal access is based on Article 3 of the Oviedo Convention and, in this case, implies the absence of harmful discrimination[11]).

(e) Provision of measures to adequately inform the population or part of the population about the existence, purposes and means of access to the screening program, as well as the voluntary nature of participation in it (individuals should be able to know about the existence of the program and its purpose, the voluntary nature of their participation in it, how to access it, the benefits and possible negative impacts (e.g. exam results stress), as well as the implications for other family members[12]).

The above conditions align with the general conditions outlined in the other chapters of the Protocol (II-VII) and are applicable at an individual level.[13] Of great importance is Chapter IV, which establishes the right to information (Article 8(1)) and genetic counselling (Article 8(2)) for persons undergoing genetic testing, their free consent to it (Article 9(1)), and the free withdrawal of consent at any time (Article 9(2)). Similarly, Chapter V defines, as specific aspects of the right to respect for private life, the protection of personal data obtained from genetic testing (Article 16(2)), the right to know (Article 16(1)), and the right not to know (Article 16(3)).

According to the drafters of the Protocol, establishing a genetic testing program requires a pilot study on a small segment of the target population, aimed at assessing the program's positive and negative effects across various levels (technical, organizational, clinical, psychological, social)[14] .

V. Specific guidelines on the selection of diseases to be tested

The Commission emphasizes the importance of establishing a clear set of criteria for selecting the diseases to be tested. According to the Commission, the specific criteria for compiling a list of genetic diseases are as follows:

1. There must be strong evidence that a gene variant (or variants) causes the disease and can be reliably detected. In other words, only genes with variants known to cause a pathological condition should be investigated, and it should be possible to confirm the diagnosis with other complementary tests.

[10] Explanatory Report to the Additional Protocol, §149.
[11] Explanatory Report to the Additional Protocol, §151.
[12] Explanatory Report to the Additional Protocol, §152.
[13] Explanatory Report to the Additional Protocol, §146.
[14] Explanatory Report to the Additional Protocol, §150.

2. A large proportion of individuals with the gene variant (or variants) are expected to experience symptoms that would have a debilitating impact on their quality of life if left undiagnosed.

3. It has been demonstrated that early or pre-symptomatic intervention for the condition results in substantially better outcomes in children than intervention after the onset of symptoms.

4. The conditions screened for are only those for which interventions are equitably accessible.

VI. Additional legal principles

According to the Commission, every newborn genetic screening program must comply with the following legal principles regarding the collection and processing of personal data:

(a) The principle of personal data protection. This principle requires the implementation of appropriate technical and institutional measures to prevent the unauthorized disclosure of collected data to third parties. Pseudonymization of biological data at the source of collection is the primary measure to ensure that, when processing is carried out by a different entity, that entity does not gain access to the identities of the newborns. At the same time, pseudonymization allows the collecting entity to know the identity of the newborns, so that it can inform the parents of the results of the genetic tests performed by the processing entity, which may reveal significant health information about the newborn. Within the framework of this principle, equally important institutional measures include the appointment of a Data Protection Officer (DPO) and, most importantly, the preparation of a Data Protection Impact Assessment (DPIA) ensuring the concrete legal commitment by the data controller to implement the appropriate protection measures.

b) The principle of proportionality in the collection and processing of personal data. It is a fact that protecting identity confidentiality through technical measures depends on the volume of data collected, and it has been shown that, even under pseudonymization conditions, large datasets increase the risk of identity disclosure by the processing entity. For this reason, strict adherence to the principle of data minimization (see GDPR Article 5(1)(c)) is essential, since it is a manifestation of the principle of proportionality. In applying these principles, the Commission particularly notes that, although nearly the entire genome is sequenced for technical reasons, it is not permissible to analyze it in relation to any phenotype, due to the high risk this poses to personal data protection. Finally, these same principles require the preparation of a list of diseases, based on current literature and entries in relevant

biological databases, for which preventive measures are appropriate, as treatment after their disease onset is generally inadequate.

In conclusion, the Commission considers that the implementation of any newborn genetic screening program must be guided by the principles of **reliability and validity principles**. This requires ensuring the credibility of the processing entity so that the validity of genetic testing results is beyond question. In this context, only state-licensed laboratories that have obtained appropriate certification should be authorized to conduct genetic testing on newborns.

VII. Proposals

In view of the above, the National Commission for Bioethics and Technoethics issues the following proposals:

1. Urging Greece to sign and ratify the Additional Protocol to the Oviedo Convention on genetic testing for health purposes (27.11.2008).
2. Preparing a specific and comprehensive research protocol prior to implementing any newborn genomic screening program, including its submission, evaluation, and approval by a competent scientific committee, and the research ethics, and deontology committees.
3. Ensuring supervision by a public authority during the implementation of each genomic screening program.
4. Obtaining a consultative opinion from the National Commission for Bioethics and Technoethics prior to making decisions on newborn population genomic screening.
5. Publicizing the newborn screening policies to be implemented and providing comprehensive information through the competent authorities in order to ensure transparency and bolster public trust.
6. Providing coordinated, systematic, and responsible information to parents during pregnancy, as well as before and after the child's birth, by a specialized health professional (clinical geneticist and genetic counsellor) to ensure proper understanding of the process and the potential outcomes of expanded newborn genomic screening.
7. Introducing legal regulations governing the consent process for each planned genomic screening program.
8. Developing a framework of guidelines to govern the selection of the newborn genetic conditions to be screened, based on the available specialized

infrastructure and the management capacities of the country's National Health System.

9. Publicizing and regularly updating the list of all conditions under consideration for screening.
10. Preparing a DPIA to evaluate the impact of collecting and processing sensitive health and genetic data.
11. Implementing, on behalf of the State, an appropriate plan and policy for managing the outcomes of genetic testing.
12. Institutionalizing and strengthening the role of specialist genetic counsellors.
13. Organizing, through the Ministry of Health, appropriate policies and the necessary health system infrastructure to address the conditions under consideration.
14. Monitoring and evaluating the impact of each planned newborn genetic screening program within the framework of the existing national newborn screening program.

Athens, 21 November 2025

ANNEX

Submitted memoranda:

- **Georgios Daskalakis,** Professor of Obstetrics and Gynaecology, Director of First Department of Obstetrics and Gynecology, 'Alexandra' General Hospital, National and Kapodistrian University of Athens, Athens, (9/6/2025)
- **Christos Kattamis**, Professor Emeritus, former Director, First Paediatric Clinic, Agia Sofia Children's Hospital (7/6/2025)
- **Antonia Charitou,** Paediatrician - Neonatologist – Intensivist, Head of the Neonatal Department & NICU at Clinic REA Maternity Hospital, President of the Hellenic Society of Perinatal Medicine (HSPM), Board Member of the Union of European Neonatal and Perinatal Societies (UENPS)
- **Efstratios Hatzixaralambous**, President of the Hellenic Federation of Rare Disease Associations, (23/06/2025)
- **Georgios Chrousos,** Professor Emeritus, former Director, First Paediatric Clinic, Agia Sofia Children's Hospital (15/6/2025)