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Portuguese Health System – Challenges in Times of Genomics

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Abstract: This article presents the Portuguese health care system that combines the National Health Service, health subsystems and private health insurance. Genomic medicine is expanding the scope of its activity, and its main challenges in pre-natal medicine and post-natal healthcare will be presented here and it will be discussed whether a private health insurance system can provide genomic medicine. As the Four "Ps" concept of Medicine is increasingly relevant, it is important to check if the private system can accommodate this evolution.

1. The Right to Health Care in Portugal

The social right to health protection shall be guaranteed by the National Health Service (*Serviço Nacional de Saúde*, SNS), according to the original version of the Constitution of the Portuguese Republic of 1976 (*Constituição da República Portuguesa*, CRP). The Minister of Social Affairs, Dr António Arnaut, created the SNS through Law No. 56/79 of 15 September 1979.[1]

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[1] Unlike Article 35 of the Charter on Fundamental Rights of the European Union (Health care) and Article 3 of the Convention on Human Rights and Biomedicine, which mentions "Equitable access to health care", the Portuguese Constitution requires that access to health

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The first Basic Health Law dates to 1990[2] and, recently, Law No. 95/2019 of 4 September 2019 has taken its place.[3] The 2019 Basic Health Law aims to reinforce the central and crucial role of the patient, reinforcing patient's rights and centrality, recognizing the importance of patient associations and their representation, as well as their importance in achieving health and recognising the importance of genomic medicine.

2. Health Insurance in Portugal

In Portugal, three different systems coexist: (1) the National Health Service (known as the SNS), (2) health subsystems (social health insurance systems for certain professional groups such as security forces, health workers, and state [ADSE] and banking [SAMS] sectors[4]) and (3) voluntary health insurance, between private parties and insurers.

There are three types of insurance contract:[5] (1) reimbursement (expenses are borne by the insured person, who can request full or partial reimbursement from the insurer), (2) assistance (managed care, where there is a network of services that the insurers themselves provide and recommend to the insured) and (3) a combination of the previous two. In the market, Low-cost Health Insurance is also available – despite being commonly referred to as a "health plan", it is a health card that offers *discounts* on certain health services, such as dental care.[6]

be effective, following the Beveridge model, with financing based on taxes and providers being predominantly public (Article 64 of the CRP).

2 Law No. 48/90 of 24 August 1990, amended by Decree-law No. 185/2002 of 20 August 2002.

3 Jorge Simões, Inês Fronteira, and Gonçalo Figueiredo Augusto, "The 2019 Health Basic Law in Portugal: Political Arguments from the Left and Right," *Health Policy* 125, no. 1 (January 2021): 1–6.

4 This is a figure originating from the Bismark Model. These are funds fed by contributions paid by their potential beneficiaries or by those who are their relatives. The contributions they make to a given fund give them and their family members access to health care from selected providers.

5 *Regime Jurídico do Contrato de Seguro* (RJCS) – Decree-law No. 72/2008 of 16 April 2008, most recently amended by Law No. 82/2023 of 29 December 2023.

6 According to the *Autoridade de Supervisão de Seguros e Fundos de Pensões* (supervisory authority of insurance services), these are products that work within a network of providers and allow the customer to access a set of health services at a lower price, without the assumption of risk by the platform managing the health plan (or health card).

The number of people holding private health insurance reached almost 3.6 million in 2023,[7] which equates to over a third of the Portuguese population. The number of those insured under health policies rose by 3.3% in the first 6 months of the COVID pandemic. In 2023, the rise in inflation meant that private health care insurance prices rose by 6.7%,[8] and are expected to continue to increase throughout 2024.[9]

Thus, access to health services varies. Around 40% of Portuguese people are covered under a public or private health subsystem or private health insurance.[10] However, this service is *additional* (complementary) to the one provided by the SNS, especially regarding the type of care provided: private hospitals and clinics are mainly sought to provide consultations with specialist doctors. This means that, in 2020, despite the increasing representation of private health care in Portugal, the bulk of health care services were provided by public hospitals. For example, 65% of all surgical interventions, 84.2% of all auxiliary diagnostic and therapeutic tests, 67% of radiological exams and 75.1% of births were all conducted in public hospitals.[11] In fact, preventive care, public health policies and major health interventions are, above all, a field of action of the National Health Service.

A closer look at the economic-financial level will provide a clearer idea of the real magnitude of the difference between the two sectors. Regarding the sale of insurance, the market increased by 8.3% in 2020, to almost 950 million euros. By comparison, the Budget (consolidated revenue) of the Ministry of Health was 12,565.4 million euros in 2021 and has reached a record budget of 15.000 million euros in 2024.

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7 "Há quase 3,6 milhões de pessoas com seguro de saúde em Portugal," *Observador*, 10 December 2023, accessed May 17, 2024, https://observador.pt/2023/12/10/ha-quase-36-milhoes-de-pessoas-com-seguro-de-saude-em-portugal/.

8 Data retrieved from EcoSeguros, "INE calcula que prémios dos seguros de saúde aumentaram 6,7% este ano," 18 December 2023, accessed May 17, 2024, https://eco.sapo.pt/2023/12/18/ine-calcula-que-premios-dos-seguros-de-saude-aumentaram-67-este-ano/.

9 Carolina Ribeiro, "Seguradoras vão aumentar os preços dos seguros de saúde," *CNNPortugal*, 10 January 2024, https://cnnportugal.iol.pt/videos/seguradoras-vao-aumentar-os-precos-dos-seguros-de-saude/659ea2210cf25f99539372f2.

10 A. Mateus et al., *Sector Privado da Saúde em Portugal, Millenium BCP e Augusto Mateus & Associados* (2017), 9.

11 *Instituto Nacional de Estatística - Estatísticas da Saúde: 2020*, INE, 2022, 25.

Nevertheless, the role of private health financing is still very relevant in Portugal, mainly due to direct payments, particularly to cover medication costs. Voluntary health insurance and other out-of-pocket costs borne by families account for 36.6% of the total health expenditure in Portugal. Compared to other OECD countries, this is a very high value[12] and has a regressive impact on society, as poor individuals spend relatively more on medication and healthcare than well-off members of society.

3. Limitations of Health Insurance Contracts

The National Health Service (SNS) is responsible for providing access to health care. Health insurance is an *additional* resource and not an *alternative*. Therefore, the principle of private autonomy and contractual freedom is firmly maintained, which has its reverse in the *pacta sunt servanda* rule (Article 406 of the Civil Code). Therefore, health insurance contracts have serious limitations in their scope of protecting the right to health.

3.1. Expenses over Insurance Ceilings

Private customers can choose between three plans with different coverages and prices, which vary depending on the options selected and the insured person's age.[13] For hospital health care, the operator offers coverage between €15,000 and €500,000. In other words, if treatments exceed these values, the insurance company is no longer responsible for paying for the services.[14] For this reason, it is common, particularly in oncology, for patients to leave hospitalization (of their own free will or compelled by the private provider) or to abandon the therapeutic plan they were carrying out relying on health insurance, and then having to be placed a possible waiting list for treatments offered universally and free of charge by the SNS.[15]

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12 OECD, "Health Spending (Indicator)," 2024, accessed May 17, 2024, https://doi.org/10.1787/8643de7e-en.

13 This information was obtained from one of the key players in the insurance market.

14 According to a Lisbon Court of Appeal decision of 7 June 2018, within the scope of a health insurance contract, the Insurer may only be responsible for covering expenses up to the maximum amount of coverage stated in the policy in question. Also, the Insurer shall not be held liable for damages to the Insured that result from non-compliance with the insurance contract. (Proc. 7983/15.4T8LSB.L1–6.).

15 André Dias Pereira et al., "White Book On Cancer Patients´ Rights: Portugal," in *White Book on Cancer Patients' Rights*, eds. Anne-Marie Duguet and André Dias Pereira

3.2. Non-renewal of the Insurance Contract

Regarding the renewal of the contract, if not stipulated otherwise, the contract is valid for one year and automatically renewed if neither party declares their desire to terminate it, through a written statement sent to the other party at least 30 days before the date on which the contract would be renewed (in accordance with Article 115, No. 1, of the RJCS). Therefore, if the insurer realizes that the risks to the insured's health have worsened – for example, because a request has been made for the reimbursement of treatment expenses for a serious illness, which could involve relevant costs – the insurance company will inform the insured that it does not intend to renew the contract. In such cases, Article 217 applies. Its protection is very weak. It only states that in the event of the non-renewal of the contract or coverage, if the risk is not proportionally covered by a subsequent insurance contract, the insurer shall not, for two subsequent years (after the event of non-renewal), refuse to cover expenses resulting from the manifested illness (or any other health care expense *related* to it) as well as expenses related to any other event that occurred throughout the term of the contract. Therefore, the insurer must cover these expenses if they were initially covered by the insurance policy and up to the maximum amount of coverage provided for in the contract and still left over on the last period of contract validity (Article 217/1). Moreover, the insurer must be informed of the illness within 30 days following the end of the contract, unless there is a reasonable impediment that justifies a delay (Article 217/2).[16]

Consequently, the insurer will only provide services until the insured capital from the last period of validity of the contract is exhausted. It is understood that, if it were not for Article 217/1, the insurer would never bear these risks. This is a specific rule for health insurance contracts. If there is no subsequent contract (or if there is, it does not have identical protection to cover that disease), the insurer is by law obliged to cover the expenses

(ONCONET SUDOE Project WP 3–3, June 2019), 2–34, https://www.onconet-sudoe.eu/media/attachments/2020/08/18/whitebook_patients-rights.pdf; and André Dias Pereira et al., eds. *Cadernos da Lex Medicinae - n.º 2 | Cancro e Direito* (Coimbra: Instituto Jurídico – Faculdade de Direito da Universidade de Coimbra, 2018). Available at: https://www.centrodedireitobiomedico.org/publica%C3%A7%C3%B5es/publica%C3%A7%C3%B5es-online/cadernos-da-lex-medicinae-n%C2%BA-2-cancro-e-direito.

16 Maria Inês Oliveira Martins, "Seguros e Doença Oncológica," in *Cadernos da Lex Medicinae*, 24.

resulting from illnesses manifested during the validity period of that insurance contract.

3.3. Limits to the Duty of Providing Information

The policyholder and the insured are obliged to declare all circumstances that may be relevant to risk assessment by the insurer before the contract is concluded. The insurance provider usually prepares a questionnaire for the prospective client, which might not mention all health issues relevant to the insured. If other circumstances relevant to the assessment of the applicant's risk exist that were not included in the questionnaire, the applicant must provide such information to the insurance provider. The insurer has the obligation (or risks incurring civil liability) to clarify to the interested party this duty of providing information and the consequences of non-compliance (Article 24 of the Law on Insurance Contracts).

Article 25 stipulates that intentional omissions or inaccuracies from the policyholder may lead to the contract being declared void by the insurance company. As clarified by Portuguese jurisprudence, "It is legitimate for the insurance company to cancel the insurance contract in cases where the insured person knowingly omitted relevant data about their health."[17]

Selling insurance is not even conceivable without standardizing the terms and conditions.[18] Article 3 of the Law on Insurance Contracts stipulates that legislation on unfair contractual terms,[19] consumer protection and distance contracts also applies to insurance contracts. The unfair contract terms regime can be invoked, to both control what is included in the contract (whether the insured understood its terms and conditions) and to assess the content of the terms (for example, a clause that stipulates a period of eight days to submit a reimbursement request). On the other

17 Évora Court of Appeal Decision of 13 July 2017, Proc. 1846/13.5TBSTR.E1; Already in 2007, the same Court stated that diabetes is likely to influence the terms of a life insurance contract, thus if when answering the questionnaire about their health, which was part of the life insurance proposal they subscribed, the insured omitted the fact they had diabetes, the contract is void according to Article 429 of the Commercial Code – Tribunal da Relação de Évora, Judgement of 25 January 2007, Proc. 10091/2006–2.

18 Maria Inês Viana de Oliveira Martins, "O controlo do conteúdo das cláusulas contratuais gerais no contrato de seguro," *Revista Thesis Juris–RTJ* 9, no. 2 (2020): 416.

19 Law on General Contractual Terms (Decree-law No. 446/85 of 25 October 1985, last updated by Decree-law No. 123/2023 of 26 December 2023).

hand, the Law on Insurance Contracts itself is cautious in relation to health insurance, considering its special risk, and imposing some requirements.

In health insurance, the insurer covers the policyholder's risks related to health care that do not correspond to the agreed benefits or the expenses incurred in each year of contract validity if a random event provided for in the same contract occurs, obliging the same policyholder to pay the corresponding price.

Pursuant to article 216 of Decree-Law No. 72/2008, pre-existing illnesses, unknown to the insured person at the date of the contract, are covered by the coverage agreed by the insurer, and may be restored by agreement to the contrary; however, the contract may provide a grace period not exceeding one year to cover these diseases.

If the contract ceases to be valid as a result of non-renewal and if the risk is not covered under the subsequent insurance contract, the insurer shall not, in the two years following the termination and until the insured capital has been exhausted in the last period of contractual validity, refuse to provide benefits resulting from illness or other health care benefits related to the term of the contract, as long as they are covered by insurance (Article 217(1)).

3.4. Health Insurance in the Basic Health Law – Reinforcement of the Duty of Information

Bearing in mind the low protection provided by health insurance, the 2019 Basic Health Law (Article 27) reinforced the duty of information setting some basic requirements:

1) subscribing to an insurance or health plan must be preceded by the insurer providing clear and intelligible information regarding the conditions of the contract, i.e. the scope, exclusions and coverage limits, including information on possible interruption or discontinuation of health care provision if the contractual insured coverage limits are reached;
2) health establishments are required to inform users about the costs of health care services when patients have subscribed to insurance and health plans. This includes informing about the costs of the entire proposed medical intervention (except when the institutions justifiably do not have the necessary elements to provide this information). The contract must therefore be *clear* about what is covered by the insurance

and what is excluded, how the contribution is made (total or partial), coverage caps, deductible values, the deadlines to submit the reimbursement request and what that process entails;

3) requirement to provide clarifications regarding the age limit criterion. Lifetime health insurance is currently excluded in Portugal, individuals must be warned that, after a certain point in their lives, they will not be able to enter a health insurance contract and, if one is in force, it will terminate without the possibility of renewal. Most insurance contracts do not allow people over 60, 65 or – in exceptional cases – over 70 years old;[20]
4) the insured must be informed that there will be an interruption or discontinuation of health care provision if the contractually established maximum limits are reached. Health establishments are also required to provide information to avoid financial "surprises" at the end of treatment or hospitalization.

3.5. The Right to Insurance after the Treatment of a Serious Illness – The "Right to Be Forgotten" – An Incomplete Legal Reform

In an effort to improve the legal level of protection of those citizens who wish to be *additionally* protected through a health insurance contract, the Parliament enacted an important Law, which unfortunately is not yet applicable due to lack of regulation.[21]

Law No. 75/2021 of 18 November 2021 introduced several changes to Law No. 72/2008.[22] It reinforces the right of people who have overcome or mitigated situations that posed an increased risk to their health or have had a disability, to access credit and insurance contracts, thus prohibiting discriminatory practices and establishing the "right to be forgotten."[23]

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20 Cf. Sofia N. Silva, *Os Seguros de Saúde Privados no contexto do Sistema de Saúde Português* (Associação Portuguesa de Seguradoras, 2009), 42.

21 Luís Poças, "A Lei 75/2021, o direito ao esquecimento e os seguros," *Revista de Direito Comercial* (2022); on this issue, see also an article: "Lei do Direito ao Esquecimento aguarda há dois anos por ser regulamentada e cumprida," Portal de Informação Português de Oncologia Pediátrica, December 13, 2023, accessed May 17, 2024, https://www.pipop.info/lei-do-direito-ao-esquecimento-aguarda-ha-dois-anos-por-ser-regulamentada-e-cumprida/.

22 Law No. 72/2008 (the Law on Insurance Contracts or RJCS) was amended in 2015, 2021 (by Law No. 75/2021) and, most recently, in 2023 (through Law No. 82/2023).

23 The previous law which prohibited discrimination based on disability or increased health risk (Law No. 46/2006 of 18 November 2006) did not include anti-discrimination rules

This right has long been claimed by survivors of pediatric cancer and other patients who have mitigated situations of increased health risk and those who overcame a disability and allows them access to various financial products, such as life insurance or bank credit, without being compelled to declare that they had an illness posing a substantial risk to their health.[24]

Discriminatory practices that violate the principle of equality are forbidden (Article 15 of the Law on Insurance Contracts). This follows the general rule *prohibiting discriminatory practices* and enforcing the principle of equality laid down in Article 13 of the Constitution. Discriminatory practices, such as charging different premiums to those with a disability or increased health risk are not allowed. Yet, as the Law is not yet regulated, health insurance is not yet adequate for the health protection of those citizens.[25]

designed to protect those who *overcame* an increased health risk or a disability. Therefore, the 2021 Law – influenced by French law – increases its scope of application by widening the concept of "increased health risk." Francisco Rodrigues Rocha, "O 'direito ao esquecimento' na lei n.º 75/2021, de 18 de Novembro: breves notas," *Revista da Faculdade de Direito da Universidade de Lisboa* 63, no. 1–2 (2022): 341–64.

24 Law No. 75/2021 prohibits discriminatory practices, enshrines the "right to be forgotten" and allows cancer patients who have overcome or mitigated situations of aggravated health risk or a disability to have, as consumers, the "right to be forgotten" when taking out loans to acquire housing or consumer credit, as well as in the contracting of mandatory or optional insurance associated with said credits. The law also ensures that they shall not be subject to an increase in the insurance premium or exclusion of guarantees from insurance contracts, and no health information relating to the medical situation that gave rise to the increased health risk or disability may be collected or processed by credit institutions or insurers in a pre-contractual context. The law imposes some requirements, namely relating to the time frame in which the treatment ended: 10 years must have passed since the end of the therapeutic protocol in the case of an increased health risk or disability that has been *overcome*; 5 years – since the end of the therapeutic protocol, if the medical condition occurred before the age of 21; or 2 years of continued and effective therapeutic protocol are required in the case of increased health risk or disability that is being *mitigated*. The Law does not aim to impose the principle of equality (where all situations are treated equally) but rather ensure that similar situations are treated similarly, and a different procedure is adopted for different situations, ensuring the observance of the principle of equity.

25 Despite having come into force on 1 January 2022 (Article 8), Law No. 75/2021 still has certain aspects relating to insurance activity that require regulation by the Government. According to Article 7 of the Law, this regulation should have been completed before 1 January 2023. This delay has motivated the Assembly of the Republic to issue a Resolution recommending that the Government should regulate to make Law No. 75/2021 operative. But, as yet (April 2024), no Regulation or Ordinance has been published.

3.6. Health Insurance Is Not an Instrument for Universal Coverage

In conclusion, Portuguese law continues to entrust the National Health System (SNS) with an increasing responsibility for health care provision. Health insurance, increasingly common in Portuguese society, still has many usage limits and is freely revocable – by the insurance company after one year. It is not, therefore, seen as a life-long "health plan" or long-term solution that protects citizens who suffer from serious and/or chronic illnesses, especially as their health concerns increase with age.

With this legal and societal framework, resulting from a certain level of legal traditionalism, no clear lines of consumer protection for people who acquire health insurance have been developed. The general principles of private law still govern many aspects of health insurance, with the additional regulation of the law on general contractual terms and conditions and sector regulation.

4. Genomic Medicine

4.1. From the Human Genome Project to the Three Ps (Predictive, Preventive and Personalised) Medicine

In the 1990s, the study[26] of topics related to genetic engineering in the field of law began, along with the establishment of ethical and legal rules.[27] Launched in October 1990 and completed in April 2003, the Human Genome Project accelerated scientific research in clinical genetics.

Nowadays, genomics is thriving, thanks to gene editing technologies such as CRISPR/Cas9 that allow the genome of any organism, including humans, to be modified in a controlled manner.[28] Moreover, in the field of human genome analysis, there have been advances in genetic tests, performed at a very reasonable cost and with a very significant level of reliability and precision. In the context of healthcare provision, genetics, combined with artificial intelligence, is opening doors, with "personalised" and

[26] Guilherme de Oliveira, "Implicações jurídicas do conhecimento do genoma," *Revista de Legislação e Jurisprudência* 129 (1996): 325–32.

[27] At the level of the Council of Europe (Convention on Human Rights and Biomedicine), and UNESCO (Universal Declaration on the Human Genome and Human Rights of 1997 and the International Declaration on Human Genetic Data of 2004).

[28] André Dias Pereira, "Gene Editing: A Challenge for Homo Sapiens," *Medicine and Law* 36, no. 4 (December 2017): 5–28.

"precision" models with clear gains in terms of effectiveness and safety of treatments opening the door to the predictive preventive and personalized medicine – 3PM.[29]

In human health,[30] the applications of genomic medicine are essentially divided into two areas: reproductive medicine and post-natal preventive and therapeutic interventions.[31] Its cost and its implications represent an ethical, legal, and economic challenge to publicly funded health care and are probably out of reach for the private insurance legal scheme in force in Portugal.

4.2. Genomics and Reproductive Medicine: Prenatal Diagnosis and Preimplantation Genetic Diagnosis

Advances in biotechnology and early diagnosis have been posing tremendous ethical challenges to the legal good of intrauterine life and are contributing to an expansion of the so-called *individual or liberal eugenics*, centered on individual choices, resulting in the transfer of the "decision-making axis from politicians to parents."[32] In this context, the legal norms should safeguard human dignity, the physical and psychological integrity of women and the protection of the embryo.

In Portugal, pre-natal tests are allowed[33] and abortion is legal up to 24 weeks of pregnancy, in case of serious fetal malformation (Article 142 Penal Code).[34]

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29 Fernando J. Regateiro et al., "Promoting Advanced Medical Services in the Framework of 3PM—a Proof-of-Concept by the 'Centro' Region of Portugal," *EPMA Journal* 15 (2024): 135–48, https://doi.org/10.1007/s13167-024-00353-9.

30 The CRISPR/Cas technique is also used in animals, plants, and environmental changes in agriculture, biofuel production, modification of endangered animal species, food characteristics, etc. – Bill Gates, "Gene Editing for Good: How CRISPR Could Transform Global Development," *Foreign Affairs* 97, no. 3 (May/June 2018): 166–70. Cf. Thaís Cesa e Silva, *A Edição Genética como elemento das responsabilidades parentais: Uma antecipação do cenário juscivilístico familiar face aos avanços da Engenharia Genética* (Coimbra: Instituto Jurídico, 2021).

31 André Dias Pereira, ed., *Medicina Personalizada de Base Genómica, Boas Práticas, Ética e Direito* (Coimbra: Coimbra University Press, 2023).

32 Jürgen Habermas, *The Future of Human Nature* (Cambridge: Polity, 2003).

33 Ordinance No. 5411/97 of the Ministry of Health.

34 Article 142 permits abortion if it is performed by a doctor and in the following scenarios: (1) abortion is the only method to avoid the risk of death or grave physical or mental harm

Concerning Preimplantation genetic diagnosis (PGD), Law 12/2006 accepts it in Article 29: for people from families with mutations that cause early death or serious illness, when there is a high risk of transmission to their offspring.[35] The National Council of Ethics – 51/CNECV/07 – stated in 2007 that:

> The use of the DGPI puts into conflict ethical values that may come into conflict in certain circumstances. When it is possible to avoid the development of a human being who has a high probability of being born with or developing a serious illness, which leads to premature death and prolonged suffering and is irreversible, the use of DGPI can be positively valued from the point of ethical view.

These ethically problematic technologies are provided both in the public and the private sectors. In the public sector, there are some difficulties in accessing PGD, since there is a long waiting list.[36] In the private sector, this very expensive procedure is paid out of pocket, as insurance companies (normally) do not offer this service.

4.3. Post-natal Genomic Medicine

This sphere of medical activity is very large and growing, and poses serious challenges for a health system. The Medicine of the Three Ps (1. Prevention 2. Prediction 3. Personalization [to which one shall add a fourth P – 4. Participation]) is one of the outcomes of Genomic Medicine. This evolution implies

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to the mother; (2) abortion is recommended in order to avoid the risk of death or permanent grave physical damage to the mother – up to the 12th week of pregnancy (3) the foetus is at risk of grave illness or malformation – up to the 24th week of pregnancy; (4) the pregnancy was caused by rape or sexual assault – up to the 16th week of pregnancy; (5) at the mother's choice – up to the 10th week of pregnancy. In cases where the foetus is not viable, abortion can be performed at any time during pregnancy. Any of the conditions mentioned above must be certified by a doctor, except item 5, in which case the mother has to submit an affidavit to a doctor or clinic stating that her decision was "mindful and responsible". If the mother is under 16 years old or mentally incapacitated, the consent to perform an abortion has to be provided by the woman's legal representative (usually parents).

35 The National Council for Assisted Reproductive Technology decides *case by case* and the medical indications for possible PGD are determined by current good practices and are included in the recommendations of national and international professional organisations in the area and are reviewed periodically.

36 See opinion 98/CNECV/2017 – Opinion on Waiting Lists When Carrying Out Pre-Implantation Genetic Diagnosis (Pgd).

the proper management of individual genomic data, a practice guided by the demands of legal and ethical principles, particularly data protection.

The advantages often attributed to personalized medicine are: (1) identifying diseases earlier (accurate diagnosis), (2) reducing treatment burdens, and (3) tailoring treatment to the patient (pharmacogenomics).

However, personalized medicine is not without its problems. The widespread sharing of personal databases and biobanks created for biomedical research, as well as the scientific results obtained, raises questions about the right to confidentiality of this information and the privacy of participants. Thus, confidentiality and privacy in the clinical context may be affected, and there may be a violation of the right not to know one's own genetic information. Moreover, it requires expensive resources and qualified personnel.

Predictive and presymptomatic genetic tests must have strong legal protection, as they contain familial information, (potentially) definitive/permanent information, and information with potential discriminatory effects. Therefore, predictive genetic information that is particularly sensitive includes: (1) Potentially *permanent* genetic information because it is (potentially) unchangeable throughout life; (2) *Familial* genetic information, as it provides a lot of information about ancestors, descendants, and about close relatives, which raises social, psychological, ethical, and legal issues; and (3) *Predictive* genetic information, as it can provide information that a certain healthy person will suffer (in the case of monogenic diseases) or with some probability will suffer (multifactorial diseases) from a certain serious disease, which can be a source of disturbance in personal, family and professional life, and in relationships with institutions, such as insurers. Since 2005, Law 12/2005 has protected people against discrimination based on genetic grounds, specifically in the context of (life and health) insurance, labor and adoption. However, in their actual practice insurance companies normally exclude patients with genetic diseases, either through the investigation of family history or after the onset of the symptoms by not renewing the insurance contract.

Insurance companies continue to violate an old law, Law 12/2005, which in Article 12 (4) states that "Insurance companies cannot demand or use genetic information obtained through the collection and registration of the client's family history as a reason to refuse the subscription of

an insurance, to charge a higher premium or for any other purposes." This is a daily practice and the competent Authorities do not intervene in a clear violation of the Law and the duty not to discriminate based on an individual's genetic profile, also established in the Constitution and the Convention on Human Rights and Biomedicine.[37]

4.4. Gene Therapy

Furthermore, genomic medicine may offer somatic gene therapies and germline gene therapies.

4.4.1. Somatic Gene Therapies

Regarding *somatic line therapies*, good news continues to emerge, offering hope for many people and families affected by severely disabling genetic diseases, causing immense physical, psychological, and social suffering. In this context, one should highlight phase 1 or 2 clinical trials that, in the last 10 years, have included the use of genomic editing (targeting in vivo or ex vivo somatic cells) for treating diseases such as sickle cell anemia, β-thalassemia, genetic blindness, some forms of cancer, diabetes, urinary tract infections, familial amyloid polyneuropathy, hereditary angioedema and HIV/AIDS.

Somatic gene therapy has been ethically and socially accepted, only requiring strict adherence to the classic rules of biomedical research, with a strong emphasis on risk-benefit decision-making. In Public Law, but also Insurance Law, it mainly raises issues related to the principles of equity and (bioethical) justice, due to the high cost of drugs produced based on it. In fact, one may wonder if such medications are affordable for private insurance companies in a market as small and unregulated as the Portuguese one.[38] On the other hand, how far can the SNS afford such high costs? So far, the Portuguese National Health System has been able to accommodate innovative therapies. In the last 5 years, over 260 innovative medicines were

[37] Article 11 – "Any form of discrimination against a person on grounds of his or her genetic heritage is prohibited."

[38] Recently, the FDA approved a drug based on gene editing that costs 3.5 million dollars – Michael Cook, "The World's Most Expensive Drug Is a Cure for Haemophilia," *BioEdge*, November 29, 2022, accessed May 17, 2024, https://bioedge.org/public_health/the-worlds-most-expensive-drug-is-a-cure-for-haemophilia/.

incorporated into clinical practice with co-participation or full payment of costs by the State.[39]

4.4.2. Germline Gene Therapy

As for germline gene therapy, its use is very controversial. It should be noted, however, that, despite the prohibitionist basis set out in Article 13 of the Convention on Human Rights and Biomedicine, drawn up in 1997, over the last few years there has been a progressive acceptance[40] of the use of germinal genetics, i.e. if the intervention is carried out with therapeutic purposes for the subject (embryo) of the therapy and their descendants (avoiding diseases such as Huntington's disease, Portuguese Familial Paramyloidosis and – in the future – even diabetes, hypertension etc.), this may prove to be acceptable, as long as the technique is safe, which is not yet the case.[41]

Concerning somatic or germline genetic editing for *enhancement* purposes, it is prohibited by international documents and international organizations.[42]

The UNESCO International Bioethics Committee, in October 2015, called on member states to accept a moratorium on modifications to the germline through genetic editing.[43]

In 2017, the National Academy of Sciences and the National Academy of Medicine of the USA stated: "Germline interventions, within strictly

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39 See: Agência Lusa, "Mais de 260 medicamentos inovadores entraram no arsenal terapêutico nacional em 5 anos," *Observador*, July 23, 2023, accessed May 17, 2024, https://observador.pt/2023/07/23/mais-de-260-medicamentos-inovadores-entraram-no-arsenal-terapeutico-nacional-em-5-anos/.

40 Portuguese Basic Health Law (Basis 11) is open to future progress, stating: "The State recognises the importance of genomics in the context of public health, and the law must regulate genomics for therapeutic purposes, carrying out tests and knowledge base of data for the provision of healthcare and research, in compliance with the following principles: (…) e) Freedom of scientific research in genomics, taking into account its importance for the improving the health of individuals and humanity."

41 See: André Dias Pereira, "Gene Editing: Portuguese Constitutional, Legal and Bioethical Framework," in *Rechtliche Aspekte der Genom-Editierung an der menschlichen Keimbahn. Veröffentlichungen des Instituts für Deutsches, Europäisches und Internationales Medizinrecht, Gesundheitsrecht und Bioethik der Universitäten Heidelberg und Mannheim*, eds. Peter Axer et al. (Berlin: Heidelberg, 2019), https://doi.org/10.1007/978-3-662-59028-7_14.

42 Eduardo António da Silva Figueiredo, *Desagrilhoar Prometeu? Direito(s), Genes e Doença(s) – Desafios Constitucionais na Era da Engenharia Genética* (Lisboa: Petrony, 2020).

43 The Council of Europe's Bioethics Committee Declaration on Genome Editing Technologies of 2 December, 2015.

regulated risk limits and when coupled with accompanying research on the risk, were ethically defensible if the intervention constituted 'really the last reasonable option' for a couple of having their own healthy, biological child." In other words, the official position on such interventions shifted from "not allowed as long as the risks have not been clarified" to "allowed if the risks can be assessed more reliably." This represents rather an extraordinary methodological and hermeneutic inversion. From prohibition, it moved to a moratorium and then to conditionality.

In September 2017, international concerns, notably expressed by the German Bioethics Committee were that "speculations now concentrate less on whether but rather only on when the first human genetically modified by genome editing will be born."[44]

In November 2018, it was announced in China that twin girls with genome modifications in the germline had been born. On 29 November 2018, the CNECV took a public stance, condemning this event.[45] In recent years, there have been no reports of other human rights violations through genetic editing of embryos.

In December 2018, the World Health Organization (WHO) established an advisory committee of global and multidisciplinary experts (the Advisory Committee on Developing Global Standards for Governance and Oversight of Human Genome Editing) to examine the scientific, ethical, social and legal challenges associated with (somatic, germline, and hereditary) human genome editing.[46]

Also, the UNESCO panel of experts calls for the prohibition of "editing" human DNA to avoid unethical manipulation of hereditary traits,

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44 Deutscher Ethikrat Berlin, 29 September 2017.

45 "Manipulação Genética em Embriões humanos através do uso de técnicas de edição de genoma," Conselho Nacional de Ética para as Ciências da Vida, November 29, 2018, accessed May 17, 2024, https://www.cnecv.pt/pt/comunicacoes/manipulaco-genetica-em-embries.

46 Expert Advisory Committee on Developing Global Standards for Governance and Oversight of Human Genome Editing; As a result of this committee's work, in 2021, WHO issued new recommendations. It emphasises the need to avoid the premature use of genetic editing in humans. See: World Health Organization, *WHO Expert Advisory Committee on Developing Global Standards for Governance and Oversight of Human Genome Editing. Human Genome Editing: Position Paper*, 2021; World Health Organization, *WHO Expert Advisory Committee on Developing Global Standards for Governance and Oversight of Human Genome Editing. Human Genome Editing: A Framework for Governance*, 2021.

stating: "Gene therapy can be a watershed in the history of medicine, and genome editing is undoubtedly one of the most promising endeavors of science for the benefit of all humanity."[47]

Will the prohibition continue to be imposed on couples who increasingly invoke procreative freedom, the "right" to have healthy offspring provided with "well-being" – arguments that can mobilize the very Declaration of the World Health Organization (WHO) from 1946, which states: "Health is a state of complete physical, mental, and social well-being and not merely the absence of disease or infirmity." Others go further and even foresee the legal duty of a family to seek healthy offspring, using technological means available in a particular society. Thus, it is also necessary to revisit Savulescu's thesis on the moral obligation of parents to choose the best possible genetic constitution for their children, which leads some authors to foresee the future fit of a parental legal obligation to resort to genetic interventions for preventive-therapeutic purposes.[48] Regarding the challenges that genetic editing poses to society and law, it is important to recall Jonas's words: "Act in such a way that the effects of your action are not destructive of the future possibility of such life."[49]

5. Conclusion

Genomic medicine brings new hopes and fantastic advancements in the struggle for a healthier life. It allows couples to embrace a reproductive project with the hope of having a healthy child, it cures severe diseases, and it makes it possible for individuals to prevent illnesses or to adopt an adequate medication and lifestyle. However, this predictive preventive personalised (and participatory) medicine requires strong and stable financing and the democratic engagement of all stakeholders, and also respect for

47 "Universal Declaration on the Human Genome and Human Rights," UNESCO, accessed May 17, 2024, https://en.unesco.org/themes/ethics-science-and-technology/human-genome-and-human-rights.

48 Julian Savulescu, "Procreative Beneficence: Why We Should Select the Best Children," *Bioethics* 15, no. 5/6 (2001): 414–26; Julian Savulescu and Guy Kahane, "The Moral Obligation to Create Children with the Best Chance of the Best Life," *Bioethics* 23, no. 5 (2009): 274–90. Silva, *A Edição Genética*.

49 Hans Jonas, *Technology, Medicine and Ethic, On the Practice of the Principle of Responsibility* (Frankfurt, 1990).

the human rights of all citizens, irrespective of age, health condition, health risks, and genetic heritage.

The National Health Service is struggling to respond to these complex challenges. On the other hand, the private insurance sector – as it is nowadays organized in Portugal – does not seem to be prepared to face these challenges with some compassion.

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