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BIOÉTICA EN GENÉTICA: ENTRE EL ARTE Y LA CIÊNCIA

Ana Rita Soares[1] *https://orcid.org/0000-0001-7817-9889*
Ana Sofia Carvalho[2] *https://orcid.org/0000-0003-1132-8880*

[1] Centro de Genética Médica Dr. Jacinto Magalhães, Centro Hospitalar Universitário de Santo António, Porto, Portugal
[2] Instituto de Ciências Biomédicas Abel Salazar, Universidade do Porto, Porto, Portugal

Ana Rita Soares - anaritamsoares@gmail.com | Ana Sofia Carvalho - anasofiapintodecarvalho@gmail.com

Corresponding Author
Ana Rita Soares
Largo da Maternidade Júlio Dinis, 20
4050-101 - Porto - Portugal
anaritamsoares@gmail.com

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EDITORIAL

BIOETHICS IN GENETICS: BETWEEN SCIENCE AND ART

Medical Genetics is a transversal and multidisciplinary medical speciality whose activity encompasses, in a general and simplistic perspective, the diagnosis of rare genetic diseases and genetic counselling of hereditary pathologies. Genetic information is, more than medical information, health information and it has implications not only for the self but also for family members. Although this medical specialization deals with risks and probabilities based on exact sciences and rigorous laboratory methodology, it also keeps challenging professionals to think ethically in different daily situations. Even in cases where the law is specific and quite rigorous on how to deal with the problem, it can be challenging to know which is the most ethical conduct in this rarity world. Henceforth, some examples will be given to make us think about how genetics can be a science art with certainties and queries.

The Genetic Information

Several studies have shown that the Portuguese population has a high level of health illiteracy and prefers to delegate health decisions to health care professionals (HCP). Even so, every day HCPs ask for patients' consent for procedures or treatments. More specifically speaking, genetic information is hard to understand even by differentiated people, and it is easy to predict how difficult it is for the general population to understand this abstract and complex information. When informed consent is given, the principle of autonomy is implicated and certain assumptions may, unintentionally, not be met albeit the information is usually given adequately and the patient decides freely and without duress, we are not entirely sure that the patient understands the information, given what was previously stated. Furthermore, processing genetic information, diseases and risks takes time, and the medical appointments are getting shorter and faster, challenging even further to explain, listen, dialogue, and understand personal, medical, or genetic information. So, are we having the most ethical and virtuous conduct with our patients?

Disclosure of Genetic Information

The Portuguese law is quite rigorous on which genetic information is medical (i.e., can be assessed by all health professionals that deal with the patient, who, by himself, presents with some disease manifestation), or health information (i.e., must only be assessed by a Medical Geneticist, as the person is healthy and presents a risk for developing or transmitting a genetic disease). Health information is not to be shared with employers or insurance and should not be assessed in the hospital records. However, most HCP are unaware of this law and, unfortunately, we see this kind of information registered in hospital records almost every day. Perhaps raising genetic education for HCP could help minimise these non-conformities. Ethically speaking, everything previously mentioned makes sense: we are protecting individual autonomy and confidentiality and providing the greatest benefit. Nevertheless, ethical issues arise when we talk about the nondisclosure of genetic information (related to severe actionable disease) to family members. This happens when a patient has (the risk of) a genetic disease, for which we have surveillance or treatment options (actionable), and he/she refuses to share this information with his/her family members who can also be at risk. If, on one hand, we want to protect individual confidentiality and autonomy, on the other hand, we should also consider the beneficence and non-maleficence of the family member at risk to have such a disease. Albeit the law protects the right to confidentiality, and this is also a perspective for normative ethics, a more utilitarian ethical perspective would do the best to the highest number of people and contact the family. Perhaps in the perspective of virtue ethics, a good dialogue and shared decision could help to shed some light on this dilemma.

Prenatal and Pre-Implantation Diagnosis

The beginning of life is a subject of ethical dilemmas and controversy since the bioethical early days, and it may not have any correct or final answer even nowadays. It would give us several pages of different perspectives, so we will give just a brief comment on genetic ethical issues.

According to Genereviews®, and regarding several diseases, "Differences in perspective may exist among medical professionals and within families regarding the use of prenatal testing (when the testing is being considered for the purpose of pregnancy termination or for early diagnosis). While most centres would consider the use of prenatal testing to be a personal decision, discussion of these issues may be helpful."

Abortion questions are classically discussed, but questions regarding termination of pregnancy for a genetic disease or malformation (according to the severity), or whether this disease would be suitable for embryo selection in preimplantation diagnosis still get some divergent opinions. Albeit this technology has started in order to help couples to have a healthy baby, without a specific rare and severe genetic disease, it can give us the "power" to choose which embryo, i.e., which live, is worth living. This capacity of "playing God", choosing which embryos can be implanted or are excluded, can take us, in extremis, to moral

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disengagement, risking the so-called slippery slope effect: instead of selecting embryos by the severe disease, the technology allows us to select embryos for sex or, going further, for the eye-colour.
These ethical issues are of concern to different HCPs and scientists, but Medical Geneticists have the moral duty to be aware of ethical principles, as well as the possible implications of their opinion and conduct.

Many other clinical situations concerning genetics could be discussed ethically. Several dilemmas, whether due to existing directives (legal or not) or due to accumulated experience, are relatively easy to solve in clinical practice. Others are becoming more and more challenging with the exponential growth of technology and genetic information, which is more and more easily and widely available.
Genetic counselling, which should be informative and not directive, requires dedication and understanding from the clinician, as each patient is unique in assimilating information and making decisions. The universal rules of Genetics become particular and personal when we apply them to a subject, a unique and unrepeatable human being, bearer of individual dignity, inserted in a society of moral values and ideals. With ethical conduct, Genetics becomes a science art, demanding justice and equity, non-maleficence, respect for the person (as an individual and as a part of a family and a society), and beneficence.