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Article

Racial bias and genomic underrepresentation in the context of polygenic risk scores in embryos: implications for genetic discrimination in assisted reproduction and the protection of vulnerable people

Vieses raciais e sub-representação genômica no contexto dos escores de risco poligênico em embriões: implicações para a discriminação genética na reprodução assistida e a proteção de pessoas vulneráveis

Sesgos raciales e infrarrepresentación genômica en el contexto de las puntuaciones de riesgo poligênico en embriones: implicaciones para la discriminación genética en la reproducción asistida y la protección de las personas vulnerables

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Abstract

Objective: To understand how the relationship between the genomic under-representation of vulnerable populations in assisted reproduction and racial biases implies dynamics that may be related to genetic discrimination and the exacerbation of inequalities. **Methodology:** This will be an analytical-descriptive, conceptual and exploratory study, using a deductive approach to carry out a conceptual review of how the genetic information of vulnerable people relates to genetic discrimination in assisted reproduction. **Results:** a) the effectiveness of polygenic risk scores in embryos has challenges in non-European populations due to the lack of representative genomic data, especially involving groups of African descent; b) an alarming absence of patents addressing genomic underrepresentation in patent filings was observed, suggesting a lack of concern for genetic diversity in assisted reproduction. **Conclusion:** Genomic under-representation and the lack of genetic diversity in assisted reproduction tend to accentuate the risk of genetic discrimination, revealing a technological privilege in favor of European and white middle-class populations.
Keywords: Race Factors; Genomic Medicine; Reproductive Techniques Assisted; Health Vulnerability; Social Discrimination.

Resumo

Objetivo: compreender como a relação entre a sub-representação genômica de populações vulneráveis na reprodução assistida e os vieses raciais implica em dinâmicas que podem estar relacionadas à discriminação genética e a exacerbação das desigualdades. **Metodologia:** utilizou-se pesquisa analítico-descritiva, com concepção conceitual e exploratória, que se utiliza do método de abordagem dedutivo para realizar revisão conceitual sobre como as informações genéticas de pessoas vulneráveis se relacionam com a discriminação genética na reprodução assistida. **Resultados:** a) a eficácia dos escores de risco poligênico em embriões tem desafios substanciais em populações não europeias devido à falta de dados genômicos representativos, especialmente envolvendo grupos

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afrodescendentes; b) observou-se uma alarmante ausência de patentes que abordem sub-representação genômica em depósitos de patentes, sugerindo falta de preocupação com diversidade genética na reprodução assistida. **Conclusão:** a sub-representação genômica e a falta de diversidade genética na reprodução assistida tendem a acentuar o risco de discriminação genética, revelando um privilégio tecnológico em favor de populações europeias e de classe média branca.
Palavras-chave: Fatores Raciais; Medicina Genômica; Técnicas de Reprodução Assistida; Vulnerabilidade em Saúde; Discriminação Social.

Resumen

Objetivo: Comprender cómo la relación entre la subrepresentación genómica de las poblaciones vulnerables en la reproducción asistida y los sesgos raciales implica dinámicas que pueden estar relacionadas con la discriminación genética y la exacerbación de las desigualdades. **Metodología:** Será un estudio analítico-descriptivo, conceptual y exploratorio, utilizando un enfoque deductivo para realizar una revisión conceptual de cómo la información genética de las personas vulnerables se relaciona con la discriminación genética en la reproducción asistida. **Resultados:** a) la eficacia de las puntuaciones de riesgo poligénico en embriones ha sido cuestionada en poblaciones no europeas debido a la falta de datos genómicos representativos, especialmente en grupos de ascendencia africana; b) se observó una alarmante ausencia de patentes que aborden la infrarrepresentación genómica en las solicitudes de patentes, lo que sugiere una falta de preocupación por la diversidad genética en la reproducción asistida. **Conclusión:** La infrarrepresentación genómica y la falta de diversidad genética en la reproducción asistida tienden a acentuar el riesgo de discriminación genética, revelando un privilegio tecnológico en favor de las poblaciones europeas y blancas de clase media.
Palabras clave: Factores Raciales; Medicina Genómica; Técnicas Reproductivas Asistidas; Vulnerabilidad en Salud; Discriminación Social.

Introduction

One of the sensitive and problematic aspects of reproductive medicine today is, according to Chapman[1] to understand how to ethically use ethnic-racial issues and genetic ancestry in reports of technological innovation in assisted reproduction without causing some kind of discrimination or stigmatization, especially in the context of vulnerable people.

Understanding that there are racial biases that need to be explored within the algorithms designed for the Polygenic Risk Score (PRS) highlights a duty of increased care, especially in sizing up how these biases can potentially misrepresent disease risk, increase health inequalities between populations and even result in stigmatization and genetic discrimination of vulnerable populations due to a lack of generalization[2].

According to Alwin *et al.*[3] (p. 13) "[...] vulnerable people are those who, for different reasons, have diminished capacities to face possible violations of basic rights, of human rights". Afro-descendants are the people who tend to be the most disadvantaged in terms of genetic discrimination and disparities in the context of access policies for the technologies analyzed in the background so far.

Article 8 of the Universal Declaration on Bioethics and Human Rights (UDBHR) regulates respect for the human vulnerability of individuals and specific groups and for individual integrity. This is because the normative command establishes that "[...] Human vulnerability must be taken into account in the application and advancement of scientific knowledge, medical practices and associated technologies".

In Brazil, the majority of the population is unable to afford most of the new technologies in assisted reproduction. In a scenario of social inequality, the protection of specific vulnerable groups

and individuals must be redoubled in the application of technological advances in reproductive medicine. Here it is important to understand that the protection of vulnerable people is not a broad perspective. It must be interpreted restrictively, under the terms of the UDBHR, since the meaning of the normative command refers to "individuals and groups of specific vulnerability" [4].

For Franklin[5], reproductive technological innovation can be seen as a coming together of "twoof the most powerful Euro-American symbols of future possibility: children and scientific progress" as a combination of "two of the most powerful Euro-American symbols of future possibility: children and scientific progress". Thinking about a prospective scenario for technological innovation in assisted reproduction also means thinking about improving or prioritizing the genetic quality of the human population [6]. Vulnerability is present in this scenario, with special reference to research involving human beings. This is because Morais and Monteiro[7] argue that there are specific groups of people who deserve differentiated treatment, taking into account that they are "[...] more exposed and less able to defend themselves from abuse and mistreatment by others". In a restrictive conception of vulnerability, Kotzé[4] explains that certain individuals "are vulnerable to harm or risk in a way that many other individuals or communities are not".

Therefore, the condition of vulnerability also affects individuals, groups of people, communities and even countries whose autonomy and capacity to deal with situations of risk and harm in the face of technological advances in assisted reproduction have been diminished[4]. However, the aforementioned author[8] states that:

> When used to identify a collective, however, it does not necessarily mean that every individual in that group is vulnerable or that vulnerability is an immutable constant. Consequently, vulnerability is also a gradual and relational notion (Ten Have, 2016: 11). It is also possible to distinguish different forms of vulnerability; while medical vulnerability usually comes to mind first in bioethical discussions, Ten Have also mentions 'physical, psychological, social, economic and environmental vulnerabilities' (2016: 11) [...] It is then also necessary to further clarify who is meant by people who have a special vulnerability. As Ten Have points out, the notion 'that people are vulnerable simply because they belong to specific groups or populations is inadequate' (2016: 75). When reference is made to vulnerable populations, it does not necessarily mean that, within a given group, all people are affected in the same way (Ten Have, 2016: 131).

In the context of medical research, Schroeder and Gefenas[9] maintain that vulnerable people can be subject to exploitation, that is, "[...] they are easily taken advantage of unfairly to serve the interests of others". Depending on the project or research in reproductive medicine, many donors of genetic material may feel compelled to donate, according to Kotzé[4] "[...] for the financial compensation and because they live in scenarios that make it unlikely that they will be in a position to use assisted reproductive technology treatments themselves, should they need or wish to. However, they donate so that others can benefit from a biotechnology to which they do not have access". This is a reality that needs to be observed in the context of "systems in which vulnerable people do not benefit" [4] because:

> [...] when poor young women from vulnerable communities in the developing world are lured into egg donation in order to be financially compensated, and the recipients of these donations are wealthy people from the developed world, this description of vulnerability rings true[4].

The prospect of conceiving a child has always been based on systemic inequalities, especially with regard to reproductive technologies. These inequalities increase the individual's vulnerability to the risk of harm[4].

This article deals precisely with the limits and possibilities of access to genetic risk information in a context of genetic discrimination and vulnerability, based on the following objective: to understand how the relationship between the genomic under-representation of vulnerable populations in assisted reproduction and racial biases implies dynamics that may be related to genetic discrimination and the exacerbation of inequalities.

To this end, the research question that guided the analysis of this article was the following: is it possible to establish a relationship between the scarcity of genetic information in vulnerable populations and the occurrence of genetic discrimination in assisted reproduction?

Methodology

This was an analytical-descriptive study, with a conceptual and exploratory approach, which used the deductive method to carry out a conceptual review of how the genetic information of vulnerable people relates to genetic discrimination within a framework that makes it possible to understand the parameters of access to risky genetic information.

This section was established by the empirical approach outlined by the findings of Rodrigues [10] in his patent investigation on the Espacenet platform, which mapped the current stage of technological development of PRS applied to human embryos. This perspective was crucial to the construction of an analytical framework based on a critical investigation into the intrinsic limitations of PRS for historically vulnerable people.

The aim was to combine knowledge of medical genetics, bioethics and health law within a transdisciplinary approach. The aim of this approach was to overcome barriers between disciplines in order to understand the scientific object in its entirety[11] (p. 59) which not only underpinned the conceptual analysis, but also the proposition of paths for future research and public policies in the area. This integrative approach made it possible to examine the challenges of genomic under-representation and the possible risks of discrimination in assisted reproduction.

Results and discussion

Several experts have been discussing the study "Genome-wide risk prediction of common diseases in human pre-implantation embryos"[12] published in the journal *Nature Medicine*, which aimed to explain how to predict the risks of some common diseases in pre-implantation fertilized embryos. In the article, the limitations of the study were described; one limitation in particular draws attention and is emphasized by Helen O'Neill [13] "[...] polygenic risk scores have limited efficacy in non-European populations due to lack of representation in genome sequencing databases".

In the field of genetic information, polygenic risk scores offer the possibility of identifying the risk of polygenic diseases in adults or even in embryos before implantation, since these diseases are responsible for a significant proportion of premature deaths in the human population.[2] In assisted reproduction, these scores can be used as part of embryo selection methods in specialized clinics to

[2] Information taken from Spacenet, patent registered as WO2022055747A1 – *PREIMPLANTATION GENETIC TESTING FOR POLYGENIC DISEASE RELATIVE RISK REDUCTION.* Disponível em: https://worldwide.espacenet.com/patent/search/family/080629804/publication/WO2022055747A1?q=WO2022055747A1&queryLang=en%3Ade%3Afr.

predict the risk of various diseases[3], such as Alzheimer's, autism, schizophrenia, and type 1 and 2 diabetes.

Some assisted reproduction clinics in Brazil already offer the polygenic risk score service for *in vitro* fertilization, and these options can be easily found on the internet. A simple Google search for the acronym "PGT-P", which refers to "pre-implantation genetic diagnosis for polygenic diseases", reveals several clinic websites that offer this type of service(10).

It is important to note that, in addition to predicting the likelihood of developing complex diseases, the potential of using polygenic risk scores (PRS) to identify complex characteristics unrelated to health, such as height, impulse control, level of intelligence or personality traits, is already being debated(14). If this technology proves to be effective and safe, concerns are raised about the permissibility of its use in cases involving both diseases and non-medical characteristics(14). This raises thoughts about the possible increase in social inequality if PRS is made selectively accessible(14).

The clinical usefulness of polygenic risk scores in embryos has yet to be proven. This is especially true given the fact that a large part of genetics is based on a Eurocentric perspective, i.e. studies of European descendants. This means, according to Helen O'Neill(13) that it is necessary to scale this limitation when diversifying embryonic genomes. According to Joyce Harper(13) "current diagnostic techniques are not available to non-European descendants, which may exacerbate the view that in vitro fertilization is seen as a white middle-class treatment".

Wang *et al.*(15) (p. 306) explain that the effectiveness of polygenic risk scores (PRS) has substantial challenges in populations considered to be admixed, "for which two or more ancestral components (usually originating from different continents) are present in each genome". These are largely under-represented populations. Treff *et al.*(16 (p. 1163) argue that this point involving greater genetic diversity between various ethnicities is a situation to be solved "[...] in existing biobanks, which is fundamental to PGT-P technology." According to Wang *et al.* there are studies that point out that the prediction performance of polygenic risk scores of European origin suffers "with increasing admixture ratios of underrepresented ancestry, especially African ancestry". By combining polygenic risk scores (PRS) of European ancestry and under-represented non-European ancestries, there is a tendency, in the views of the authors mentioned(15) to improve the prediction of diseases of mixed ancestry.

An aggravating factor is that November *et al.* (2) (p. 2097) note that underrepresented and marginalized groups "[...] suffer from environmental health disparities, such as exposure to pollutants, nutritional deficits or lack of drinking water", which can exacerbate the challenges within the gene-environment interaction to be reflected in the applicability of PRS.

It's interesting to note that November *et al.*(2) (p. 2096) that:

> [...] future development of PGS incorporates greater cohort diversity across multiple dimensions (e.g., ancestry, age, gender, access to health care, and other environmental variables relevant to focal phenotypes). Specifically, funding agencies and researchers should aim to form more diverse cohorts and make the best use of these resources through collaborative data sharing. In recent years, several such efforts, including the

[3] "Disease risk' refers to the probability that an existing person or a person born by *in intro* fertilization of an embryo will express a specific disease based on an interpretation of genetic data informed by empirical data or bionformatic modeling. "Information taken from Spacenet, patent registered as WO2022055747A1 – *PREIMPLANTATION GENETIC TESTING FOR POLYGENIC DISEASE RELATIVE RISK REDUCTION.* Disponível em: https://worldwide.espacenet.com/patent/search/family/080629804/publication/WO2022055747A1?q=WO2022055747A1&queryLang=en%3Ade%3Afr.

> *All of Us, H3Africa and Trans-Omics for Precision Medicine (TOPMed)* programs (see Web Resources), have been launched with the aim of increasing the diversity and representation of previously understudied groups in human genomics research and better understanding the contribution of genetic and environmental factors to disease risks.

According to Forzano *et al*.[17] (p. 493):

> The estimation of PRS for children of parents from diverse ethnic backgrounds is not yet possible to determine correctly. In order for risks to be calculated as accurately as possible, PRS must be combined with the effects of an individual's non-genetic factors, such as life history, environment, nutrition and physical activity.

Martin *et al*.[18] (p. 584) argue that the biggest ethical and scientific debate regarding the clinical application of PRS, a technique that is commercially available worldwide, including in Brazil, is that "today they are several times more accurate in individuals of European descent than in non-Europeans". It represents the aforementioned Eurocentric biases in genome-wide association studies.

There are efforts, albeit measured, to improve the diversification of populations in order to remedy imbalances. This is currently the most critical limitation of precision medicine genetics, which is highly dependent on population representation[18]. The verification of the nuances of this imbalance, in the authors' view, has already been demonstrated theoretically, based on "[...] simulations and empirical bases on traits and diseases"[18] (p. 585). The point is:

> [...] the poor ability of genetic studies to generalize across populations stems from the overwhelming abundance of studies of European ancestry and the paucity of well-founded studies in globally diverse populations. This imbalance is especially problematic because previous studies have shown that studies on Hispanic/Latino and African-American individuals contribute an outsized number of associations relative to similarly sized studies on Europeans. More worryingly, the fraction of non-European individuals in genome wide association studies has stagnated or declined since late 2014 [...] thus suggesting the absence of a trajectory to correct this imbalance [18] (p. 584-585).

It is important to consider that "most PRS methods do not explicitly address recent admixture [...] thus, further methodological development is required" [18] (p. 585). This statement converges with the content of the patent applications analyzed in the recent study by Rodrigues[10] on the state of the art of polygenic risk scores in human embryos in the *Espacenet* database, since only one patent analyzed explicitly mentioned the challenges of reproductive technological innovation involving miscegenation. Even so, the reference to miscegenation in the aforementioned patent filing entitled *Multi-gene risk score for in vitro fertilization*[4] was intended to reveal that polygenic risk, in some respects, is unable to explain miscegenation.

This means that ethnic-racial issues and genetic ancestry, as described in the patent applications analyzed by the aforementioned author, do not seem to be a concern in the patent field.

However, a crucial point that requires an interdisciplinary approach, including the participation of legal experts, in the evolution of polygenic risk scores (PRS), is the ethical issue related to the use

[4] Available from: ttps://worldwide.espacenet.com/patent/search/family/075338602/publication/CN114728069A?q=CN114728069A.

of ethnic-racial and ancestry data in PRS reports in assisted reproduction, with the aim of avoiding any kind of discrimination or stigmatization[1].

A disturbing issue that deserves attention when examining the patents analyzed by Rodrigues[10] is the fact that, of the seven eligible applications, at least four[5] prioritized or even relied exclusively on population genotype data obtained from tissue samples from the *UK Biobank* for the construction of disease predictors. The *UK Biobank* is an extensive biomedical database that gathers detailed genetic and health information from half a million UK participants, considering both genetic predisposition and environmental exposure in the development of diseases[19].

As Marshall teaches [20] (p. 251-256):

> Emerging technologies in machine learning and artificial intelligence will make it possible to predict a person's risk for these complex traits. Advanced algorithms developed using sophisticated statistical models help researchers understand how genes interact with each other to influence polygenic traits [...]. To determine the relevant genes and their appropriate weights, computers use PRS-specific *software* to analyze genome-wide association studies ("GWASs"). The data for GWASs comes from large biobanks containing hundreds of thousands of individuals′ complete genomic data that are publicly available through projects such as the UK *Biobank* and the US *All of Us Research Program.* GWASs allow researchers to identify potential genetic markers for a specific trait through statistical analysis by comparing two cohorts: a cohort of people affected by a trait and a similar cohort of people without that trait. The algorithms compare the complete genome of each study participant with that of all the other participants in search of genes that appear in the affected cohort at a higher rate than in the unaffected cohort. The more genetic variants the algorithm takes into account, the stronger the predictive power of the final PRS algorithm.

The fact is that genome-wide association studies often face the problem of ascertainment bias. In a comparative analysis of the sociodemographic health characteristics of the *UK Biobank* participants and the general UK population, it was found that the participants were healthier, wealthier and "94.6% of them were of white ethnicity, which was similar to the UK national population"[19] (p. 1027). This reveals a clear lack of population genetic diversity.

According to Wang *et al.*[15] the clinical usefulness of polygenic risk scores (PRS) is not yet definitive, nor is it clear or widely applicable. The authors argue that the accuracy of PRS varies between different populations due to the biases of predominantly Eurocentric genetic studies. The crux of the problem lies in the fact that assessing the accuracy of PRS depends heavily on the context and diversity of population ancestry. Assessing the predictive value of PRS for admixed individuals (with diverse origins) is a challenge both in terms of data and methods, which can result in the overestimation or underestimation of disease risk in certain populations[15].

Genome-wide association studies should be easily accessible to diverse populations, even though underrepresented or marginalized populations are a smaller fraction of the size of the European dataset in the specialized literature[18] and in international patent filings, with white Europeans predominating in these databases.

If the data extracted is applied to other populations, this can, in Laura Hercher's view[21] be considered misleading. Dealing with genetic data involving miscegenation represents a precaution that must be implemented in conjunction with ethnic-racial issues in the genomic field. This is because,

[5] Patents 3, 4, 8 and 9 in Annex I of Rodrigues' article (10).

according to the researcher[21] "[...] it's population data, which means you have to be very careful about how you use it for an individual. Specifically, it means that the clinical use of polygenic risk scores, at the present time, can exacerbate health disparities if measures are not adopted to address ethnic-racial issues. In this sense:

> The clinical use and deployment of genetic risk scores must be informed by the issues surrounding tests that would currently unequivocally provide far greater benefits to the subset of the world's population that is already on the favored side of health disparities[18] (p. 587).

Afro-descendant populations, in the view of Martin *et al.*[18] (p. 587-588) are:

> [...] less likely to benefit from improvements in PRS precision health care delivery with existing data, due to human population history and study biases. This phenomenon is a major concern globally and especially in the United States, which already leads other middle- and high-income countries in both real and perceived economic and health disparities.

In a scenario of genetic discrimination in technological innovation in assisted reproduction, regulatory protections are necessary to delimit the parameters of clinical trials involving population genetic diversity. Although there are bills in Brazil that deal with genetic discrimination in different areas, it is in the United States that there are rules aimed at curbing this concept in the context of health insurance and employment opportunities through the Genetic Information Nondiscrimination Act of 2008[22].

Therefore, thinking about the normative dimension is also particularly concerned with the protection of minorities and marginalized groups in the context of innovation in precision medicine. However, Martin *et al.*[18] point out that a drastic change in priority is needed, given that the process of genetic diversification between studies involving different populations has remained stagnant in recent years.[6] This is because

> [...] the growing interest and scale of genetic studies in low- and middle-income countries (LMICs) raises ethical and logistical considerations about data generation, access, sharing, security and analysis, as well as clinical implementation, to ensure that these advances do not occur. They don't just benefit high-income countries. Structures such as H3ABioNet, a pan-African bioinformatics network designed to enable H3Africa researchers to analyze their data in Africa, provide cost-effective examples for training local scientists in LMICs[18] (p. 589).

This means that in order to avoid genetic discrimination in technological innovation in assisted reproduction, there needs to be an increase in genetic diversity in genomic analysis. In this respect:

[6] "[...] Global efforts [...] link genetic data, clinical records, and national registry data across more homogeneous continental ancestries, such as UKBB, BBJ, China Kadoorie Biobank, and Nordic efforts (for example, in Danish, Estonian, Finnish, and other integrated biobanks). Notably, some of these biobanks, such as UKBB, have participants with considerable global genetic diversity, allowing for multi-ancestry comparisons; although minorities in this cohort provide the largest deeply phenotyped GWAS cohorts for various ancestries, these individuals are frequently excluded from current statistical analyses in favor of the simplicity offered by analyzing only the largest genetically homogeneous European ancestry data. [...] The most notable investment here comes from the Human Heredity and Health in Africa initiative (H3Africa), increasing genomic research capacity in Africa through more than US$ 216 million in funding from the US National Institutes of Health and the Wellcome Trust (United Kingdom) for genetic research led by African investigators." [18] (p. 589).

> Several large-scale publicly funded datasets exist, such as the *Million Veterans Project* and *Trans-Omics for Precision Medicine* (TOPMed), but with data access problems where even GWAS summary data within and between populations are not publicly shared[18] (p. 589).

Therefore, there is a latent need to promote policies for the responsible sharing of genomic data[1,23,24,25]. Otherwise, "American and European associations fear the emergence of a genetic under class"[26] (p. 138). One of the challenges of these genomic data sharing policies to overcome is the potential for stigma and discrimination when acquiring, storing and using genetic data and information in inappropriate and abusive ways in biobanks[1,27].

Conclusion

We elucidated that polygenic risk scores in embryos have limited efficacy in non-European populations due to the lack of representation of genome sequencing data for different populations. In this scenario, populations of African descent and mixed-race populations are largely under-represented. This means that there are racial biases that should be investigated within the context of embryo polygenic risk score algorithms to prevent stigmatization and genetic discrimination of vulnerable populations.

One alarming point is the lack of documents exploring any kind of concern about racial bias, miscegenation and under-representation, especially in patent filings on the *Espacenet* website.

In this context, systemic inequalities are part of the assisted reproduction landscape worldwide, making vulnerable people prone to undue exploitation in the interests of third parties.

In the scenario of technological innovation in assisted reproduction, genetic information is abundant for European descendants, but not for non-European populations. This exacerbates genetic inequalities in terms of the effectiveness of polygenic risk scores for different populations and appears to be a scenario of privileged technological access for certain people, especially those from the white middle class.

Therefore, it is important to focus on genetic information that combines polygenic risk scores for European ancestry and diverse populations, seeking to methodologically develop approaches for this purpose. At this point, it is suggested that further research is needed due to the complexity of the subject, especially when initiatives such as that developed by the Global Alliance for Genomics and Health serve as a parameter, by promoting policies aimed at drawing up guidelines, standards and orientations for the ethical sharing of genomic data[1.28].

Conflict of interest
The author declares that there is no conflict of interest.

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