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Chromosomal disorders and Bioethics

Cromosomopatías y Bioética

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Abstract:

Human beings have approximately 30,000 genes, distributed in 23 pairs of chromosomes, giving a total of 46 chromosomes of which 23 correspond to each parent. During the process of cell division, both physical and gametogenesis, spontaneous or induced accidents may occur that produce chromosomal anomalies or chromosome disorders that result from a greater or a lesser amount of hereditary material, and are the cause of some syndromes such as Down syndrome (trisomy twenty-one), but also of spontaneous abortions and diseases or problems in growth and development. In recent years, techniques have been developed to detect chromosomal anomalies from a DNA sample that does not turn out to be non-traumatic for the mother or baby. Because of this reason, prevention and diagnosis of chromosome disorders have gained great relevance in maternal and child health care. Nonetheless, these types of procedures have created ethical policies and legislation under the responsibility of paternal action on the fetus, in the presence of a chromosomal alteration that can culminate in disability or fetal or neonatal death. For this reason, health workers must seek to provide genetic counseling that guides parents in making decisions following their ideology, culture, and socioeconomic level, without leaving aside the respect for the bioethical principles of the fetus, which is why the development of an international consensus that directs the implementation of genetic counseling is essential.

Keywords:

Chromosomopathies, bioethics, genetic counseling.

Resumen:

El ser humano tiene aproximadamente 30.000 genes, distribuidos en 23 pares de cromosomas, dando un total de 46 cromosomas de los cuales 23 corresponden a cada progenitor. Durante los procesos de división celular, tanto somática como en la gametogénesis, pueden ocurrir accidentes espontáneos o inducidos que producen anomalías cromosómicas o cromosomopatías que resultan de una cantidad mayor o menor de material hereditario y son causa de algunos síndromes como síndrome de Down (trisomía 21), pero también de abortos espontáneos y enfermedades o problemas en el crecimiento y desarrollo. En los últimos años, se han desarrollado técnicas con las que pueden lograr detectarse anomalías cromosómicas, a partir de una muestra de DNA que no resulta ser no traumática para la madre o el bebé. Razón por la que la prevención y diagnóstico de las cromosomopatías, ha cobrado gran relevancia en el cuidado de la salud materno-infantil. No obstante, este tipo de procedimientos han creado polémicas éticas y de legislación en virtud de la responsabilidad de acción paterna sobre el feto, ante la presencia de una alteración cromosómica que pueda culminar en discapacidad o muerte fetal o neonatal. Es por ello, que el personal médico debe buscar brindar asesoramiento genético que oriente a los padres a la toma de decisiones de acuerdo con su ideología, cultura y nivel socioeconómico, sin dejar de lado el respeto a los

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principios bioéticos del feto, razón por la que resulta fundamental el desarrollo de consensos internacionales que dirijan la implementación del asesoramiento genético.

***Palabras Clave**: Cromosopatias, Bioéica , Asesoramiento genético.*

INTRODUCTION

Chromosome is the system where DNA is contained, keeping its structure and integrity with the help of other molecules. It is found in the cell nucleus, and its function is to allow the transmission of genetic information to descendants randomly (1). Humans have approximately 30,000 genes distributed in 23 pairs of chromosomes, constituting a tola of 46 chromosomes, from which 23 correspond to each parent. There are two types of chromosomes, the autosomes, which are the same in men as in women, and the sexual chromosomes, having a chromosomic constitution of man 46, XY, and for women 46, XX (2).

Most chromosomic alterations are produced from the damage to the ovum or sperm causing modifications in the fragments of genetic material, this might vary a lot in some cases than in others, resulting in both an increase and the loss of chromosomes (3). Chromosome disorders are diseases that terminate in a greater or lesser quantity of genetic material and cause congenital anomalies in live newborns between 0.7 and 1.5%, being the cause of developmental morphological anomalies, requiring special medical attention (4).

In recent years, techniques have been developed that might detect chromosomal anomalies from the sample taken from DNA, neither is traumatic for the mother nor the baby (5). Fetal medicine or any other branch of medicine has preventive actions, diagnosis, and treatment, together with the application of specialized tests that allow the fetus as a sensitive patient to receive the necessary attention (6). During the last 30 years of evolution, this specialty has achieved a level of complexity that authorizes effective prenatal care based on the mother's identification of risk factors (7).

The management of the affected patient requires going beyond the diagnosis and the medical aspects, this implicates the family and implies addressing the psychological impact of the diagnosis, with the quality of the family's life(8). According to these considerations, the effect of malformation and the treatment regarding the quality of life framework need to be evaluated. It is a global concept that contemplates all the areas of interest and includes factors defined by objective and subjective indicators, both the perception and evaluation of each person concerning his well-being (8).

The objective of genetic studies is to evaluate the risk of transmitting or developing diseases, that in some cases can lead to damage in psychological levels affecting self-esteem. These bioethical conflicts arise from decision-making based on the results obtained from the mentioned studies (9).

The patient generally suffers a condition that produces sensations of vulnerability or loss of control about future events. Likewise, there are modifications of how others think about the patient and their perception about themselves, which means, how they perceive (10). Because of that, studies for detecting chromosomal disorders must have as basis willingness as a couple to do them and be informed about the actions taken about their health (9).

The main purpose of this research work is to identify and analyze the information about subjects of chromosomal disorders and their relation to bioethics, referring to aspects such as: What is it known about the topic? What features have affected individuals? How does bioethics influence the diagnosis of chromosomal disorders?

METHODOLOGY

For the search for information, PUBMED was used to gather reliable sources related to chromosomal disorders and bioethics, concerning scientific articles, medical magazine articles, digital libraries, and scientific gazettes that guarantee the truthfulness and credibility of the information described in this work.

CHROMOSOMAL DISORDERS

Each chromosome harbors thousands of genes responsible for the formation of proteins involved in the development, growth, and chemical reactions in the body. Nonetheless, during the first stages of the fetus' development, certain types of chromosomal anomalies can be developed, they are classified as numerical or structural (5). The alterations in the number of chromosomes (aneuploidies and polyploidies) are defined as the variation in the number of haploid chromosomes (n=23).

On the contrary, polyploidy is the addiction to one or more haploid complements (3n, 4n…, etc.).The addiction or loss of chromosomes results in a non-multiplot number of 23 known as aneuploidy. The profits of one or two homologous chromosomes are named trisomy and tetrasomy. Meanwhile, the loss of a chromosome is known as monosomy (5). A balanced structural alteration shown in any of the parents (carrier) does not have health effects; however, it is possible to transfer an unbalanced chromosomal complement to the offspring to the descendants, which can cause gestational losses, infertility, or even born live products with multiple morphological anomalies (11).

Corresponding to the chromosome anomalies are causes of diverse conditions. Such is the case of Down syndrome (trisomy 21) spontaneous abortions, and diseases or problems with growth and development. As a result, these chromosomal anomalies cause disease (5).

Because of this, prevention and diagnosis of chromosomal disorders have been relevant in maternal and child health care. Based on the practice of non-invasive techniques such as ultrasound, chorionic villus biopsy, and amniocentesis (7). Nevertheless, these types of procedures have created ethical and legislative controversies under the responsibility of the father's action concerning the fetus, in the presence of a chromosomic alteration that can lead to a fetal or neonatal disability or death (12).

BIOETHICS

It is imperative to state that bioethics is a systematic study of the moral dimensions including moral vision, decisions, behaviors, and policies of the sciences of life and health care through implementing various ethical methodologies using an interdisciplinary context (13).

Therefore, bioethics as a discipline intends to make an ethical reflection on the problems related to life, the reason why it has had an impressive boom in the field of fetal medicine and prenatal diagnosis. The topic has been believed not only by medical professionals but also by lawyers, philosophers, theologists, and governments (13).

Bioethics within perinatology has looked for deeming the fetuses as person-patient over parents do not violate their principles of autonomy, beneficence, and maleficence, even though they do not have the faculty to decide about themselves. Due to the above, physicians are the ones who must contemplate the risks of the necessary techniques for both the diagnosis and the treatment, departing from establishing an independent judgment that considers the best interest of the patient and allows parents to be involved in decision-making (12).

Based on the previously mentioned, bioethics seeks a complex analysis of ethical issues that might cause the development of perinatology, so its objective will be:

- Highlighting the importance of prenatal monitoring as part of the strategies for promotion and prevention of health.
- Preventing maternal-fetus death.
- Offering therapies in favor of the fetus that avoids as far as possible postnatal complications.
- Counseling the mother for the evolution and prognosis of the pregnancy.
- Averting damages in the mother and the fetus during the prenatal attention process.
- Transmitting reasonable hope to parents with the suited treatment.
- Supporting timely and appropriate decision-making in case of trouble (12).

Genetic Counseling

Genetic counseling is a process of communication regarding human troubles associated with the occurrence or risk of recurrence of a genetic disorder in a family, the objective is to help the individual and the family to:

- Comprehending diagnosis, likely course of the disease, and available management.
- It will reflect on how inherited factors contribute to diseases and the risk of recurrence.
- Understanding alternatives and options to manage risk.
- Choosing an action course appropriate for the family depends on their risks, family objectives, and ethical and religious principles (14).

As stated by the World Health Organization (WHO) includes guidelines in respect of " all individuals must have the right to know his genetic and descendant risks, and must be educated about these risks and have the reachable option of a safe termination of the gestation in case of affected fetuses or whether future parents desire it".

Notwithstanding, this proposal did not obtain complete agreement due to ethical conflicts with medical genetics about the chosen termination of pregnancy after counseling, assisted reproduction, and the status of the human embryo for research (15).

Because of the foregoing, genetic counseling must contemplate the beliefs and cultures of the families who ask for it, recognizing two types of guidance: management and non-management.In the case of guidance, management refers to the therapeutical approach from the medical personnel and how this could include their own religious, political, and eugenic ideas about parents' decision-making.

Corresponding to the non-management approach is based on providing all the available information about the convenient options to address the situation, supporting parents in decision-making that suits better their ideology and ethical principles. As a consequence, diverse studies discredit management consulting, owing to parents' decisions about genetic diagnosis, which can be influenced by the medical personnel's beliefs and considerations (15).

Genetic Diagnosis and Ethical concerns

There are distinct types of genetic diagnosis based on its purpose. For instance, predictive genetic diagnosis has as its objective to detect mutation whose aim is to foresee the predisposition of a concrete disease before appearing. As a result, the patient is asymptomatic, but his family
medical history shows evidence of developing a genetic disease.

Even so, there is no sign of a concrete moment of occurrence, the severity degree, or the first symptoms. This type of diagnosis permits taking preventive measures that can lead to reducing the extent of a disease and in some cases, even preventing its recurrence (16).

As a predictive diagnosis, we can find presymptomatic genetic diagnosis performed on asymptomatic patients whose ancestry reveals any late-onset inheritable genetic disease. In consequence, the person under study has not shown any symptoms, although it has the probability of being the carrier of a pathogenic mutation. A positive result of this diagnosis is that the individual will develop a disease.

Likeliness and risk of occurrence are so elevated that most cases, the disease is confirmed; however, the moment, the severity, and the first symptoms to manifest can be ignored (17).

The predictive genetic diagnosis also finds susceptibility genetic diagnosis. In this type of diagnosis, there will always be higher uncertainty than that of a presymptomatic. It is possible that even though mutation is detected, it is a predisposition to suffer the disease, or this does not develop. In other words, the presence of mutation increases the possibility of the occurrence of pathology,but it does not ensure it.

A positive result can help to expand the patient's monitoring and meliorate preventive measures. Moreover, the absence of mutation does not ensure the status of an individual's health (17).

A choice of predictive diagnosis is the preimplantation genetic diagnosis (PGD), a procedure that implements in vitro fertilization, followed by the extraction of one or two cells before transferring the embryos to the uterus to perform a genetic diagnosis and displace only those who are free of the studied condition. Consequently, this method becomes ideal for couples who have distinct reasons to refuse the termination of gestation in any way.

Ethical dilemmas related to PGD are mainly the moral status of the embryo and in vitro fertilization since it supposes the "production" of more embryos than the ones relocated afterward, being a problem about the treatment that the non-transferred embryos must receive. The different approaches that have been taken to the embryo and its status can be summarized in one of three ways:

1. The embryo does not have any moral status, hence the mother has the right to decide about her embryo or any other part of her body, with the same ethical considerations.
2. The embryo must have the same moral status that a human being has. After fertilization, a unique genotype can evolve into a human being. From this point of view, the embryo has its rights, and the mother's interests are not relevant to the embryo. The embryo must have the same moral status as a human being. Since fertilization, a unique genotype is established and evolves into a human being.
3. The embryo is a potential human and must be managed with dignity because it has rights that must be appreciated by the parents and society in the decision-making process (14).

Back to the previous point, the difference in the legal aspects of the situation is noteworthy. While in some countries, there are specific laws that let research with embryonic stem cells, in others, the use of human embryos is banned to obtain stem cells; nevertheless, the importation of those cells is allowed in specific cases which are not stated clearly, or the stem cells research has been forbidden since it does not have specific legislation (18). Another relevant dilemma is the access to institutions that provide the mentioned services. In countries where there are differences among social status and health services are private or depend on health insurance, there is no guarantee of access by all the members of the society. Thus pregnant women with more economic resources have better chances of performing the evaluation and prenatal diagnosis procedures.

When this happens, the principle of justice does not have equity in the distribution of the service offered as long as all the pregnant women have a specific genetic risk and have the same right to be included in the program of prenatal diagnosis. Cuba is an example of equality because all pregnant women, besides those from their cultural or social level, have cosigned access to prenatal diagnosis services, whose expenses are fully covered by the State (14).

The market for genetic tests given to the consumer has quickly grown. In many countries, regulations have not adapted to this fast evolution, which leads to unprotected consumers. In any case, these tests must fulfill settled requirements and guarantee validity and clinical utility, data confidentiality, and honest and complete genetic guidance before and after the genetic test (17).

Informed Consent

During genetic guidance and before offering the method of prenatal diagnosis, the problem and possible solutions are described to the patient. In this sense, informed consent is ” the explanation to an attentive and mentally competent patient of the nature of his illness, as well as the balance between the effects of the disease and the risks and benefits of the recommended diagnostic and therapeutic procedures, and then requesting his approval to undergo those procedures”:

1. Informed consent must have the following aspects :
2. Description of the recommended treatments and procedures.
3. Informing about the risks and benefits of the procedure.
4. Different alternatives include other treatments or procedures. In addition to risks and benefits.
5. In case of not performing the procedure, the possible results
6. Possibilities of success and what physician interprets as success.
7. Possible issues during recovery and the time in which the patient will not be able to restart her daily activities (17).

CONCLUSION

In conclusion, chromosome disorders clearly state the consequences and disorders caused not only on the fetus but also on the mother. It is necessary to think over the ethical aspects related to the way of informing the diagnosed parents about the condition, being sensible and emphatic at the moment of doing it, and as well as, notifying them about all the possible cares and considerations that must be taken into account when these types of situations happened.

Due to the psychological impact that diagnosis might have on parents’ decision-making, it might change their criterion, hence full responsibility of the parents is to provide a good quality of health to the fetus, serving his needs together with the health professionals. For this reason, medical personnel must supply non-directive genetic counseling to ensure minimal religious, political, and eugenic influences from the physician to parents’ decision-making.

On the other hand, it would be valuable if all the countries had bioethics awareness courses to sensitize the personnel, resulting in improvements in clinical, psychological, and social practices. Furthermore, all nations must have universal medical coverage, assuring access to all medical procedures that the population requires, alongside predictive genetic diagnosis, even eluding births of patients with genetic alterations. Raising awareness in the population of the importance of predictive diagnoses. It lets us see that many details need to be polished about bioethics in many countries.

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