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Genetic counseling

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Abstract

Objective: The objective of this review of genetic counseling (GC) is to describe the current concepts and philosophical and ethical principles accepted by the great majority of countries and recommended by the World Health Organization, the stages of the process, its results and the psychological impact that a genetic disease has on a family.

Sources: The concepts presented are based on an historical synthesis of the literature on GC since the 1930s until today, and the articles cited represent the most important research published which today provides the foundation for the theory and practice of GC.

Summary of the findings: The modern definition of GC is a process of communication that deals with the human problems related with the occurrence of a genetic disease in a family. It is of fundamental importance that health professionals are aware of the psychological aspects triggered by genetic diseases and the ways in which these can be managed. In the field of human and medical genetics we are still living in a phase in which technical and scientific aspects predominate, with little emphasis on the study of emotional reactions and people's processes of adaptation to these diseases, which leads to clients having a low level of understanding of the events that have taken place, with negative consequences for family life and for society.

Conclusions: The review concludes by discussing the need to refer families with genetic diseases for GC and the need for professionals working in this area to invest more in humanizing care and developing non-directive psychological GC techniques.

J Pediatr (Rio J). 2008;84(4 Suppl):S20-26: Genetic counseling, genetic diseases, primary prevention, psychological counseling, ethics and genetics, eugenics.

The origins, objectives and philosophical basis

The issue of how human societies dealt/deal with undesirable human characteristics, such as mental deficiencies and congenital malformations, has passed through the development of three basic models: the eugenic model, the preventative model and the psychological model.

The eugenic model was the first to develop. The term *eugenia* (derived from the Greek, "well-born") was initially proposed in 1885 by Francis Galton, an English mathematician and statistician who was first cousin to Charles Darwin. When positive, it would be to use science to improve desirable human qualities, such as intelligence, good physical and mental health, etc.; and when negative, to reduce undesirable characteristics, such as criminality, mental deficiencies and physical defects, alcoholism and drug abuse, etc. Even though Mendel's laws had been rediscovered in 1900 and the pioneering work by Garrod on the inheritance of certain metabolic diseases was in 1908, during the first 3 decades of the 20th century the study of human genetics was dominated by the eugenic movement, primarily represented by two institutions, The Eugenics Record Office, New York, and The Francis Galton Laboratory for National Eugenics, London. They caused great damage to the development of human genetics as a science and were politico-social movements that provided the basis for the German Eugenic Law (1933). These laws were based on simplistic deductions working from Mendelian principles, claiming, for example, that criminality, mental deficiency, etc, were determined by dominant alleles and all that was needed was to imprison those affected during their reproductive years or compulsorily to sterilize them to eliminate these "perversions" in a single generation. They promoted the establishment of sperm banks stocked by Nobel laureates,

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encouraging artificial insemination and even "stud farms for the production of Aryan children". The results were disastrous, leading to a major revolt in the scientific community, led by Penrose,[1] and eugenics was banned from humans genetics. In counterpoint to eugenics, the American biologist Sheldon C. Reed[2] proposed the term genetic counseling to describe caring for families/people with genetic diseases and argued that this care should be based on professional neutrality with relation to clients' decisions.

Principles of the eugenic model: a) the objective was to promote *eugenia*, i.e., "improvement of the human race" ; b) the use of directive and even coercive techniques: this is a State model, translated into laws based on a "rationality and responsibility" of human behavior and on giving priority to defense of the social prerogative of protection of the human "genetic pool" over the needs or desires of individuals; c) major social and political actions: compulsory sterilization, obligatory abortions, policies restricting marriage, including interracial and migratory.

Today, the major concern of the scientific unity is the return of eugenic principles as adopted by China in its Law on Maternal and Infant Health Care of 27/10/94, which mandates the following: that physicians recommend postponement of marriage if one member of the couple has active mental disease or infectious or contagious disease; if one member of the couple has a serious genetic disease, the couple must only marry if they agree to use long term contraception or to be sterilized; if a fetus is identified as having a severe hereditary disease or deformity, the physician must recommend abortion and the patient must follow the recommendation. There was, once more, intense criticism from the international scientific community about this new eugenics.[3,4] More impressive still is work published by Mao & Wertz[5] and Mao,[6] who surveyed the opinions of 255 Chinese geneticists and found that 89% of them agreed with these laws, considering that the opinion of the Chinese state was more important than individual liberty; and that 86% preferred directive genetic counseling (GC) and more than 50% agreed that genetic information should be revealed even without patient or counselee permission and even to employers or insurance companies.

The preventative, or medical model is the phase of medicalization of GC. Genetics has undergone a different development from other basic sciences of medicine, which developed within medical schools. Genetics initially developed among zoologists and botanists and it was they who perceived its relevance to human beings. As the philosophical basis of medicine widened to include the concept of prevention (preventative medicine) and with the decline in the number of deaths due to infectious diseases and the relative increase in constitutional diseases in developed countries, medicine became more and more appropriate for the development of genetics.[7] The first medical genetics departments began to appear at medical schools during the 1950s. Principles of the preventative or medical model: a) Objectives: to reduce or eliminate genetic diseases; b) genetics services come to be located at major medical centers, and genetics becomes a clinical discipline; c) the basis of GC is an accurate medical diagnosis; d) physicians become the legitimate providers of GC; e) the physician makes use of the traditional doctor-patient relationship for GC and bases their actions on the principle of neutrality (proposed by Reed[2]), however, physicians have difficulty with this model because they are more accustomed to employing the traditional doctor-patient relationship models: 1) the acute disease model: active-passive – where the doctor prescribes the conduct and the patient submits passively; 2) the chronic disease model: leadership-cooperation – where the doctor, as leader, prescribes and the patient cooperates by taking the medication.

With respect to the psychological, psychotherapeutic or psychosocial model, there are many justifications for using it in GC, including: physicians perceive that the information provided in GC are not neutral from a psychological point of view, but threaten the ego; the occurrence of a genetic disease in a family triggers off a process of mourning or suffering; people will have to deal with the emotions that are released as a result. On the other hand, physicians also perceive that other psychological situations, such as marital and sexual dysfunctions, personality types, interpersonal difficulties, religious convictions, etc., interfere in the GC process. This model recognizes that the process of GC does not only involve future reproductive decisions, but also how to deal with what has already taken place,[8] i.e., how people should be helped to adjust to their own or a family member's disease or with the risks of occurrence/recurrence, and that this is a complex psychosocial process of adjustment to (coping with) countless variables, such as previous experience with the disease, personality, motivation, education, values, culture, family and interpersonal and family dynamics. This model uses the knowledge accrued by non-directive psychological counseling (based on the person-centered approach of (C.Rogers) and employing a counselor-client relationship of mutual participation, in which counselees are helped by counselors to help themselves, in other words, to be psychologically active people.[9] This model is based on knowledge of several complex psychological processes, such as the process of building up empathy, knowledge of the processes of human suffering and mourning (its phases and management), knowledge of superficial and deep human communication (meta-message), knowledge of the processes of mechanisms of self-defense and of self-image and of the processes of decision and their relation to personality types.[9] There are great difficulties with implementing this model in practice, since professionals feel more comfortable dealing with the medical and genetic features of their patients (or clients?), and patients consult geneticists more with relation to diagnosis, prognosis, treatment or risk of recurrence than in search of help with the psychological impact of the genetic condition, risks or test results.

In truth, these different philosophies or guidelines are not mutually exclusive.[8] What cannot be accepted, and what modern human genetics does not accept, is eugenic actions in which the State exerts its force over individual liberty. The general guidance is that non-directive psychological counseling techniques should be used. In other words, clients are stimulated to be psychologically active. Nevertheless, there are situations in which experienced counselors will feel that they can support the couple by being more directive, using their experience and assurance from similar situations they have experienced.

Research led by Dorothy Wertz[10-12] has demonstrated that in all countries in the world there is a greater acceptance by genetic counselors of their patients' autonomy and acceptance of non-directive GC, although in some of the work published by Wertz[11] evidence was found for the survival of eugenic practices, even without State coercion, and for which, according to the human genetics committee of World Health Organization (WHO) there is no justification.[13,14]

General ethical considerations in genetic counseling

This section is based on what is contained in the document produced by the WHO Human Genetics Programme, published in 1998, and involving 14 countries.

The primary objective of this document was to protect people and families with genetic diseases, with the intention of assisting or provide guidance to governments and health services in member states to ensure that genetic information and genetics services are made more widely accessible in medical practice within acceptable ethical principles. The secondary objective is to allay fears and reassure the public that adequate controls are in existence to prevent abuses of genetic information and to avoid unacceptable practices, recognizing that advances in genetics have the potential to significantly improve the health of people when appropriately applied at the individual, family and community levels. This information must be integrated in an ethical manner and with respect for religious and cultural diversity.

In this document the WHO declares its preference for non-directive GC, which should be based on two basic elements: a) the provision of precise, complete and unbiased information to allow individuals to take their own decisions; b) establishment of an empathetic relationship with a high degree of understanding, so that people are helped to work towards taking their own decisions.

Below are listed the ethical principles applicable to GC:[14]

1. Respect for people and families, including the whole truth, respect for people's self determination and precise and unbiased information (autonomy).
2. Preservation of the family integrity (autonomy, non-maleficence).
3. Complete disclosure to individuals and families of all information relevant to health (autonomy, non-maleficence).
4. Protection of the confidentiality of individuals and families against unjustified intrusions on the part of employers, insurance companies and schools (non-maleficence).
5. Telling individuals they have an ethical obligation to inform their relatives that they may be at genetic risk (non-maleficence).
6. Telling individuals that it is necessary for them to reveal their carrier status to spouses/partners if the couple wish to have children and about the possibilities that their relationship may be damaged by these revelations (non-maleficence).
7. Telling people they have a moral obligation to reveal their genetic status if it may affect public safety (non-maleficence).
8. Presentation of information in as unbiased a manner as possible (autonomy).
9. Use of non-directive techniques, except with relation to treatment questions (autonomy, beneficence).
10. Involving children and adolescents as much as possible in the decisions that affect them (autonomy).
11. Obligation of health services to follow-up carriers/families if appropriate and desired (autonomy, beneficence and non-maleficence).

The current conception of genetic counseling

This concept was developed at a seminar held by the National Genetics Foundation, Inc. and published by Fraser in 1974.[15] It was then approved by the American Society of Human Genetics - ASHG in 1975[16] and became the classical conception of GC: a communication process, which deals with the human problems associated with the occurrence, or the risk of occurrence, of a genetic disorder in a family.

This process involves an attempt by one or more appropriately trained persons to help the individual or family:

- comprehend the medical facts, including the diagnosis, the probable course of the disorder (prognosis), and the available management (treatment);
- appreciate the way heredity contributes to the disorder, and the risk of recurrence in specified relatives;
- understand the options for dealing with risk of recurrence, with relation to the family's reproductive life;
- choose the actions which seem appropriate to them in view of their risk and their family goals and act in accordance with that decision;

- make the best possible adjustment to the disorder in an affected family member and/or to the risk of recurrence of that disorder.[16]

This definition emphasizes the nature of GC as being much more than a process of medical diagnosis of a clinical condition and the definition of genetic risk, and that genetic counselors should work as facilitators of a complex process of understanding what is happening to the family and act to ensure that counselees go through a process of adjustment to the new situation faced by the family: having one or more members affected by a genetic disease. Genetic diseases cause anguish or torment: miscarriages, stillbirths and neonatal mortality, very often recurrent; congenital defects of a physical (malformations), mental (mental deficiency) or sensory nature (congenital deafness and blindness), psychoses, familial cancers or diseases of specific organs or systems that generally limit quality or length of life. Therefore, a genetic disease is everything that nobody wants to discover in themselves or their family and demands a great deal of dedication from professionals, who generally should be working in multidisciplinary and interdisciplinary teams to be able to correctly manage the entire situation generated by these diseases within families and society.

Clinical situations in children where genetic counseling is necessary

Without doubt, pediatricians are the physicians who will most often come into contact with patients and families in need of genetic assessment during their professional lives and so they must always be alert to this need, particularly when presented with:

- Cases of stillbirth and neonatal mortality, which should always have their etiology defined in order that we can determine whether there is a probability of recurrence in the same family. Autopsies should be requested and pediatricians should work with families to help them to understand the need for this. The autopsy should be carried out by a team trained to identify the true etiology and prepared to diagnose genetic diseases, whether dysmorphologic or metabolic.
- Children with congenital defects, particularly when the child presents with multiple congenital anomalies.
- Children who are severely ill and where there is suspicion of genetic metabolic diseases.
- Children with genital ambiguity or abnormal sexual development.
- Children with neuropsychomotor delay or mental deficiency of undefined cause or obviously genetic etiology.
- Children with deficient or excessive growth of undefined cause or with suspected genetic disease.
- Children with specific genetic diseases of an organic system (for example: sickle-cell anemia, muscular dystrophies, etc.).

Phases of the genetic counseling process

Genetic counseling is an integrated process which should be continuous. However, there may be times at which it can be divided into phases to enable better understanding. Our description will go into greater detail about the human aspects of the process than into the technical aspects of diagnosis or of calculating risks of recurrence.

In order to better develop the subject, we will concentrate on the situation that is directly related to pediatric care, i.e., the management of couples who have had children born with severe genetic problems and which is, without doubt, the most common situation observed during consultations at genetic clinics.

The first phase: definition and/or confirmation of diagnosis

In the form of a clinical activity within the healthcare sector of Brazil, genetics is approaching its 40th birthday and has therefore spread to the country's major cities and it is now more and more common for patients to be referred to consult a genetic specialist or take a genetic test. However, this type of referral or even spontaneous demand triggers feelings of insecurity and fear. The classical genetic consultation is with the parents, who are clients or counselees because of problems with their children. The genetic counselors should initially evaluate the parents' understanding of the reasons for referral and their psychological condition, in order to truly develop empathy, i.e., so that the council can understand the emotions surrounding these people in relation to a possible deviation from normality.

Before anything else, the counselor must be adequately trained and know that:

1. Genetic diseases are different from non-genetic diseases. They are innate (part of the individual, of the self), they will never go away and, the majority, have complex prognoses.
2. The occurrence of these diseases in a family leads, when the parents become aware of it, to a process of acute suffering or mourning,[17] which can pass through several phases (shock, denial, sadness and anger, equilibrium and reorganization), which intermingle with each other and where the cycle of coping depends on many factors and much professional help and the manner in which professionals deal with their clients. The council must know how to identify the different discourses and their meanings and how to interact with these people.

It is important to clarify, as far as possible, all of the initial doubts that are manifest and explain all of the procedures

which will be performed, aiming to offer as much security as possible to the counselees.

Of course, as a consultation progresses all of the clinical procedures used in medicine will come into play, since clinical genetics has a very specific clinical methodology and work up, such morphological clinical exam with detailed anthropometric measurements, genealogy studies and specific laboratory tests (karyotype, DNA tests, specific biochemical tests), which sometimes must also be carried out on parents or other family members and the reasons for which must always be very well explained.

In the medical or preventative model, GC is based on accurate diagnosis, which is the basis for continuation of the process, however, with current levels of knowledge and technology, clinical and etiologic diagnoses could be not defined immediately and depend on the course the condition takes, which generates further feelings of fear and anguish which must be managed by the counselor.

The second phase: calculating genetic risk

This phase is more theoretical, away from contact with the family, and is the process by which professionals establish, based on the etiologic diagnosis, the causes of the disease, for example, environmental, genetic, multifactorial or unknown. If the cause is genetic, they will determine whether chromosomal or genic and, for each of these, their subtypes. Based on this etiologic diagnosis, they explain the occurrence of that particular affected child within the family, the reason for the disease, the mechanism that generated it and what the risks are of the disease recurring within the family. We will not go into details about the processes of calculating these risks, since this in itself is an extensive subject which must be dealt with separately.

The third phase: communication

Once the clinical and etiologic diagnoses have been defined and/or confirmed and genetic risk has been calculated, GC enters the phase of interacting with the family to help it to understand what has happened, i.e., communicating the medical facts (diagnosis, treatment, prognosis, cause of the disease) relating to the child or patient and the genetic risks, i.e., what is the probability that another affected individual will be born to the same family.

When should GC be carried out? I am of the opinion that genetics services should integrate themselves more and more with the institutions and teams that care for the disabled and that GC should be performed by the entire team and in a continuous manner as the case progresses and the facts indicate it is needed.

What should be said? It is part of ethical principles to tell the entire truth, however the team should reflect about to whom and in what manner it should be told, since, as we have already explained, this information is always threatening to the ego and can trigger severe emotional reactions, which need to be well evaluated and managed.

There are many problems which can interfere with communication, such as educational, linguistic and social barriers, the feelings of guilt, sustained feelings of rage and rebelliousness against professionals, marital dysfunctions, etc.

And communicating the risks? The counselor should have already determined whether there is a prior conception of the risk and whether this conception is correct or not; concepts of high or low risk should not be used to explain the risk, but their exact values should be given, exploiting understanding of the probability involved in games of chance and gambling in the explanation; expounding both sides of the risk, i.e., the chance of health (a normal child being born) and the chance of disease. It is not sufficient to do only an explanation of the risks, but it is necessary to explore the counselees' perception of the risk and the impact of that should be explored –the risk is always perceived in binary form: it may or may not happen, I can or cannot have more children. Never say, "you should not have more children ", because this would be unethical.

The fourth phase: decision and action

The family will need to take many decisions from this point on, but these will occur throughout life in response to family dynamics, and so GC should be continuous. Decisions about how to care for the child/patient; about other genetic tests; about the couple's reproductive life; about relationships and maintenance of the family and the mental health of its members; reproductive decisions and contraceptive methods; about the use of prenatal or preimplantation diagnosis or; about termination or not of pregnancies; etc.

During this phase the counselor needs to help clients or counselees to:

1. Be psychologically active, making them strive to arrive at their own decisions, accepting the risks of the consequences. Exercise the essence of non-directive GC.

2. Provide couples with knowledge about human decision-making processes: work with ambiguous feelings (demonstrate that they are normal and part of the decision-making method that human beings use, i.e., balancing pros and cons until a decision is taken, and that ambiguous feelings will stay with the person until they feel secure about the results of the decisions), interpersonal conflicts, the difficulties of decision-making and its relationships with personality types (for example: obsessive types who run away from the decision attempting to involve themselves with many other activities - hiding themselves in work; and hysterical types who hide from decisions in their fantasies).[9]

The fifth phase: follow-up

Since we recommend that GC should be continuous and should be offered at educational institutions for people with special needs, professionals should follow up families' lives more effectively and not lose contact with them, which is what happens very often nowadays. This, indeed, is what has led to a situation we are observing today: a generation of affected children born during the 1970s and 1980s and seen at Brazilian genetic services are now adults, and many have mental disabilities and are having children without being offered any care, and many of these children are also affected. The information has been forgotten and the life of the family carries on without guidance. Therefore, genetics services need to invest more in activities on a primary care level (and not just tertiary) creating conditions for more effective follow-up of the lives of families, at least those with the most significant risks of recurrence. Esmer et al.,[18] evaluated follow-up at services in the United States and concluded that it is very difficult to maintain contact with these patients and that new strategies should be planned to resolve this situation. Other health professionals are also dedicating themselves to GC, such as nursing professionals and, in some countries, a new profession of genetic counselor is being created. Much research into improving GC at the primary level has been described[19] and, even at the tertiary level, this is a true necessity. A society dedicated to genetic nursing was recently founded in Brazil and we hope it will evolve as rapidly as possible and that we can create the profession of genetic counselor.

Evaluation of the results of genetic counseling

Primarily during the 1970s and 1980s, much work was undertaken to identify what the results of the GC process were at different services around the world. This research always came up against methodological issues and its conclusions were always very much criticized. Nowadays this type of research has undergone a resurgence, particularly linked to new areas where GC is being implemented, which are the genetics of cancer and the genetics of late onset diseases, together with what are known as predictive tests.

Pina-Neto & Petean[20] published extensive work evaluating the GC process in a Brazilian public hospital, demonstrating very similar results to those reported in other locations and countries: a) the great majority of families seen at genetic services (74%) are referred there and do not know what type of service they are going to be given; b) the level of understanding (or recollection) of information is low, with 48.7% of people considered to have inadequate understanding. Just 34.9% knew the numerical risk of recurrence, while the majority knew whether the risk was high or low; c) there was a clear correlation between degree of understanding of the information and socioeconomic status; d) a large number of couples decided not to have children, irrespective of the risk of recurrence (64.4%), although couples at high risk decided not to have children with greater frequency (85.7%) than those at low risk (52.9%); the most important factor in the decision not to have children was a perceived high risk, even among those who in fact had low risk; e) there were significant differences in the use of contraceptive methods related to couples who decided to have or not to have children and related to the risks of recurrence; there was double the rate of fallopian tube ligature among high risk couples when compared with low risk couples, and vasectomy only occurred among high risk couples; f) high risk couples were more accepting of terminating pregnancies than low risk couples and men more than women; g) there was a low birth rate among all families, but the rate among high risk families was lower (five children born to 36 families –rate of 0.13, with variation in the literature of 0.24 to 0.67) than among low risk families (35 children born to 69 families –rate of 0.50, with 0.40 to 0.84 in the literature); h) recurrence in subsequent offspring was 44.4% among high risk families and 0% among low risk families; i) we had a low rate of adoption, with just three children adopted by two high risk families and just two couples split up out of the 113 who were studied (1.7%).

Conclusions and recommendations

All of the families of children diagnosed with genetic diseases must be made aware of the importance of going through the GC process. It is very important that the referring physician tries to explain as well as possible why they are making a referral and the nature of the service that will be offered. The issue of explaining the etiological mechanisms of genetic diseases, evaluating family members and defining risks of occurrence/recurrence are of great importance to families with these types of diseases.

It is fundamental that the GC process be based on non-directive counseling carried out by qualified professionals and that it be a continuous process.

Genetics services need to invest more in GC itself and in follow-up of families. We must balance technological and scientific questions with humanization of care. It is of fundamental importance to work to improve the communication process between GC professionals and families, so that the latter may obtain a better understanding of the situation they are living through and participate actively in all of the decisions they will have to take during their lives as a result of having genetic abnormalities in their families.

References

1. Penrose LS. On the interaction of heredity and environment in the study of human genetics (with special reference to mongolism imbecility).J Genet. 1932;25:407-22.
2. Reed SC. A short history of human genetics in the USA. Am J Med Genet. 1979;3:281-95.
3. American Society of Human Genetics, Board of Directors. Eugenics and the misuse of genetic information to restrict reproductive freedom: ASHG statement. Am J Hum Genet. 1999; 64:335-8.

4. Canadian College of Medical Genetics. China's eugenics law: position statement of the Canadian College of Medical Geneticists. J Med Genet. 1997;34:960.

5. Mao X, Wertz DC. China's genetic services providers' attitudes towards several ethical issues: a cross-cultural survey. Clin Genet. 1997;52:100-9.

6. Mao X. Chinese geneticists' views of ethical issues in genetic testing and screening: evidence for eugenics in China. Am J Hum Genet. 1998;63:688-95.

7. Porter IH. Evolution of genetic counseling in America. In: Lubs HA, Cruz F, editors. Genetic counseling. New York: Raven Press; 1977.

8. Walker AP. Genetic counseling. In: Rimoin DL, Connor JM, Pyeritz RE, Korf BR, editors. Emery and Rimoin's principles and practice of medical genetics. Philadelphia: Churchill Livingstone-Elsevier; 2007. p. 717-46.

9. Kessler S. Genetic counseling. Psychological dimensions. New York: Academic Press; 1979.

10. Wertz DC, Fletcher JC. Attitudes of genetic counselors: a multinational survey. Am J Hum Genet. 1988;42:592-600.

11. Wertz DC. Eugenics is alive and well: a survey of genetic professionals around the world. Sci Context. 1998;11:493-510.

12. Wertz DC, Fletcher JC, Nippert I, Wolff G, Ayme S. In focus. Has patient autonomy gone to far? Geneticists' views in 36 nations. Am J Bioeth. 2002;2:W21.

13. World Health Organization. WHO genetic counseling. Third report of the WHO Expert Committee on Human Genetics. Geneva: WHO; 1969. (World Health Organization Technical Report Series No. 416.)

14. World Health Organization. WHO ethics in medical genetics: proposed international guidelines on ethical issues in medical genetics and genetic services. Geneva: WHO; 1998.

15. Fraser FC. Genetic counseling. Am J Hum Genet. 1974;26: 636-61.

16. Mc Kusick V (President, Ad Hoc Committee on Genetic Counseling – American Society of Human Genetics – ASHG): Genetic counseling. Am J Hum Genet. 1975;27:240-2.

17. Irvin NA, Kennell JH, Klaus MH. The father of a baby with congenital malformations. In: Warkany J, editor. Congenital malformations: notes and comments. Chicago: Year Book Medical Publishers; 1971.

18. Esmer C, Urraca N, Carnevale A, Del Castillo V. Patient follow-up is a major problem at genetic clinics. Am J Med Genet A. 2004; 125:162-6.

19. Westwood G, Pickering RM, Latter S, Lucassen A, Little P, Karen Temple I. Feasibility and acceptability of providing nurse counselor genetics clinics in primary care. J Adv Nurs. 2006;53: 591-604.

20. Pina-Neto JM, Petean EB. Genetic counseling follow-up: a retrospective study with a quantitative approach. Genet Mol Biol. 1999;22:295-307.

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