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PSYCHOLOGICAL ASPECTS OF GENETIC COUNSELING FOR SENSORINEURAL CONGENITAL HEARING LOSS – DEMYSTIFYING PARENTS' BELIEFS ABOUT THE CAUSE OF THEIR CHILDREN'S DEAFNESS

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Abstract: Recent advances in molecular genetics have led to the identification of more than 400 genes that can be responsible for the etiology of congenital deafness. That means that now, genetic counseling services are available and genetic testing can be applied to certain families in order to clarify the cause of the deafness. However, in Romania, where the most common mutations are GJB2/GJB6 and W24X, we encounter a predominantly negative attitude towards genetics and its impact on deaf patients. In order to obtain better therapeutic results, studies must take into consideration the impact of genetic counseling on parental knowledge, attitudes and beliefs about genetic testing. Geneticists must also consider that some deaf parents may prefer to have deaf children and may consider the use of genetic technology to achieve this. Any genetic counselling service set up for families with deafness can only be effective and appropriate if clinicians and counsellors take into consideration the beliefs and values of the deaf community at large.[19] *Because Connexin-related deafness is common, there has been considerable discussion about introducing GJB2 and GJB6 testing into the newborn hearing screening process.*[20]

Keywords: congenital, deafness, sensorineural, genetic counseling, psychology

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INTRODUCTION

There have been rapid advances in the molecular genetics of deafness in recent years, especially after the decoding of the human genome and more than 400 different genes causing non-syndromic and syndromic deafness have been discovered. It is therefore likely that diagnostic, carrier, and, possibly, prenatal genetic testing for deafness-causing genes will become part of routine clinical practice. Uptake of such testing will depend on the understanding and opinions of the populations for whom the tests are relevant.[15] Genetic testing is a sensitive subject amongst parents as they tend to have different views and beliefs about genetics, primarily because deafness can be viewed from different perspectives. These perspectives are not readily defined in Romania but are very clear for the deaf community in Western Europe. People who refer to themselves as culturally Deaf (written with an uppercase "D") view deafness from the cultural or sociological perspective—that deafness is a condition to be understood and preserved—as opposed to the medical perspective—that deafness is a pathology to be treated or cured.[3] The deaf (lowercase "d") community is a global category for all people with any level of hearing loss, including people who are hard of hearing or deafened and deaf people who identify primarily with the hearing world as well as with the Deaf culture. Many culturally Deaf people are positive and proud to be Deaf; they have their own language (British sign language [BSL] in the UK and American sign language [ASL] in the United States) and share a common history, social customs, and identity.[3] Culturally

Deaf people are often sensitive to threats to their community; this reaction has been demonstrated clearly in the resistance to cochlear implants.[12] However, we must state that during the last 20 years views have changed dramatically and cochlear implants have become the therapy of choice for many congenital hearing loss patients. Older studies from Israel have shown that some deaf parents have said that they will not seek genetic counselling because they worry that they will be told not to have children.[17] The fear of genetic research is deeply rooted in Deaf culture, primarily because of the appalling way in which deaf people have been treated throughout history, often in the name of eugenics.[4] More recent studies have shown an improvement and a predominantly positive attitude among the public, toward genetic testing.[26]

At the present time there is no clear attitude of Romanian parents towards genetic testing and counselling, mainly due to the scarcity of these tests in the Romanian healthcare system. Psychologists, especially those who treat congenital deaf children or their families, should nonetheless take into consideration the impact of such testing on their work.

Several questionnaire-based studies could be carried out, in which parents could answer very important questions concerning their attitude towards genetic testing, such as: whether genetic testing will do more harm than good, preference for having deaf or hearing children (note that deaf parents may prefer deaf children), feelings about new discoveries in genetics, whether genetic testing devalues deaf people etc.

GENETIC CONSIDERATIONS

The human genome is formed of approximately 35.000 different genes, grouped within 23 pairs of chromosomes, 22 pairs of autosomal chromosomes and 1 pair of sexual chromosomes. Within each pair, one chromosome is inherited from the mother and the other from the father. Since the chromosomes are in pairs, genes are also in pairs.[22]

A mutation is a change in gene sequence of the DNA, compared to major population. Some mutations have no effect on the individual's health whilst others have major impact on the function of a gene

A dominant mutation – means that only one gene is necessary for an individual to be affected. Of the two copies of the gene, one from a healthy parent and the other from the affected parent, the latter will produce proteins more efficiently, in other words, will be stronger than the healthy gene and will determine the disease in the offspring. Thus, 50% of all children will inherit the chromosome with the dominant mutation.[22]

A recessive mutation - the altered gene is not strong enough to produce disease on its own and the individual must inherit altered genes (allele) both from mother and father in order to develop the disease. Individuals with only one altered gene are called carriers. These do not develop the disease but can pass the gene to the offspring. Each child has a 50% chance of receiving the mutated gene from each parent. Since in this case both genes must be mutated, the chances of an affected offspring are 25%. [22]

X-linked mutation – implies a recessive mutation for the genes on X chromosome. Since women have two X chromosomes, they will be carriers and men, who inherit one X chromosome from the mother, will develop the disease. Thus, 50% of all male offspring will inherit the mutated gene from the mother and will be afflicted. [22]

Mitochondrial transmitted mutation – represents an additional form of genetic transmission. Mitochondria are cellular structures involved in producing energy. They have a set of approximately 37 genes, different from the rest. The offspring will receive mitochondria from the mother, thus the transmission will be maternal to offspring of both sexes. [22]

Sensorineural deafness is the most common sensorial disability in humans and the genetic factor is paramount in its research and diagnosis. Until now, more than 130 loci have been identified and it is estimated that more than 100 mutations of the Connexin 26 genes are involved.[2] Genetic diagnosis is very difficult due to the variety of mutations involved. Also, the genetic involvement varies dramatically within different populations.

Recent studies indicate the mutations of genes GJB2 and GJB6 on the 13q11-q12 chromosome as responsible for more than 50% of all non-syndromic autosomal recessive congenital deafness within certain populations. These genes are responsible for encoding cellular membrane proteins such as Connexin 26 (Cx26) and Connexin 30 (Cx30) which insure the cochlear (inner ear) homeostasis. There are over 100 known allelic variants for gene GJB2, especially for autosomal recessive forms. The deletion of a single guanine (35delG) is responsible for 50% of cases of sensorineural non-syndromic deafness cases in Europe, North America and Asia.[21,23,26] The common deletion 342-Kb on the GJB6 gene (known as GJB6-D13S1830) appears in up to 20% of all deaf individuals in the U.S.A.[10,13,23]

35delG

This mutation remains the main cause of genetic deafness within the Caucasian population, with an incidence that varies from 1/35 in south-European groups to 1/79 in north-European populations [9,11] and a maximum incidence in countries around the Mediterranean.[2,7,17] This mutation is also one of the most frequent in humans and can associate other mutations of the GJB2 gene and of the GJB6 gene. Some populations such as Japanese, Chinese, Ashkenazi Jews and Rroma the mutation is rare and usually replaced by others such 235delC, 167delT or W24X.[14]

W24X

This mutation has the effect of cutting the Connexin 26 protein (24 amino acids instead of 26). It is also relatively frequent, especially in populations from northern and southern [2,18,24] Some studies from Slovakia show a high incidence within Rroma populations (26,1%), probably correlated to the Indian origin of these ethnic groups.

PSYCHOLOGICAL CONSIDERATIONS

In order to discuss the psychological impact of parents' misconceptions about the counseling for sensorineural congenital hearing loss, we should first understand the background of these beliefs.

Psychologist Daniel David in his work "Psychology of the Romanian People"[8] stated that the psycho-cultural profile of Romanians is dominanted by the mistrust of people, making them less tolerant and less cooperative when confronted to a common benefit. Mistrust can easily turn into suspicion, so Romanians seem to take a step back when faced to something new, something daring and different to every day patterns. In the same study, the author affirms that Romanians aren't a religious people in a dogmatic way, because they succeed in combining religion with pseudo-science, with heathen traditions and with all sorts of local rituals. They don't comply with the same rigors not even in respect with the church, each geographical area having its own particularities. This mistrust foundation created on the basis of a pseudo-religiosity makes the attitude towards illness be viewed in the same fantastical-mystical manner. Therefore, the core of the majority of misconceptions about illness is made by ideas like: divine punishment, parents' sin, pregnant woman who have done something wrong during his pregnancy (interruption), curses, etc.

Such beliefs make people quite passive and accept the illness as a given, manifesting a kind of passive assumption, in the idea of a sacrifice to be paid. They are not necessarily looking for solutions, and often the first interventions they make are at the level of some old anile remedies that most of the time do more harm.

So talking about genetic testing seems to be far away. In fact, even in other countries, the situation is not very good. In a 2010 study by Abe et al., "What do patients with hereditary deafness think of genetic studies?"[1], more than half (64%) of the respondents who received the genetic testing for hearing loss were not aware that it can be caused by a genetic factor. However, there is still little information about the interest in such testing. Not surprisingly, deaf adults had a predominantly negative attitude toward genetic testing for deafness, with the majority stating that they believed that such tests would do more harm than good. The majority (90%–95%) of deaf children are born to parents with normal hearing (National Information Center for Children and Youth with Disabilities 1998; Deaf World Web 2000). Inevitably, these parents search for a reason for this unexpected event and often arrive at an inaccurate conclusion[16]. In some cases, genetic testing could provide an etiology for the child's deafness, but not for all. In addition, genetic testing may provide information to assist parents of deaf children in making informed decisions concerning medical management and appropriate educational interventions for their children.[6]

Adults avoid genetics and this suggests that peoples' misconceptions and misinformation can cause some individuals to be so fearful of modern medical technology that they would decline any opportunity for genetic testing. We believe that they avoid this not because they have a misconception or a misinformation about modern technology, but because they don't have one. Moreover, a genetic test makes you somehow responsible for the child's problem, and this is not very comfortable for a parent. It is hard to accept that you are the one who caused the child's suffering, so it is preferable to blame another external cause.

In conclusion, this is clear evidence of the importance of formal pre- and post-test genetic counseling and the priority of counseling must be the educational objective. People need to know that not a curse is the cause of the hearing problem, and to understand what medical options they have.

CONCLUSIONS

In the western medical world, many studies have evaluated the beliefs and attitudes of parents about genetic testing of children with deafness. In Romania, unfortunately, no published study has specifically assessed the beliefs of parents regarding the deafness of their children and their knowledge of genetics after genetic counselling offered by professionals with specialized training. Most of parents do not know the etiology of their children's deafness until after receiving genetic counselling. Thus, genetic counselling seems to be extremely important for parents to help them understand the cause of their children's deafness, the chance of recurrence for their family members, and the mode of inheritance.

More than half of the parents are not aware that genetics can be the cause of congenital deafness and most of them have a poor understanding of the risk of having another deaf child and their deaf child's chance of having future deaf children. Authors have reported that deaf adults had a predominatly negative attitude towards genetic testing and more than half of the individuals thought genetic testing to be more harmfull than beneficial. On the other hand, the majority of hearing parents has an overall positive attitude towards genetic testing and couselling.

Genetic counselling provides appropriate information regarding genetics to patients and/or their family members to allow the clients to have their own preferred life plans. Furthermore, a genetic counsellor needs to give moral support to the clients while maintaining a favorable and confidential relationship with them. From our point of view, detecting the genetic cause of the deafness is extremely useful for diagnosis and prognosis of the illness and also for establishing its repetition rate preventive and therapeutic measures and genetic counselling.

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