Working together
www.rcis.ro

Revista de cercetare și intervenție socială

ISSN: 1583-3410 (print), ISSN: 1584-5397 (electronic)

Selected by coverage in Social Sciences Citation Index, ISI databases

GENETIC SCREENING OF DEAF CHILDREN: ETHICAL CONSIDERATIONS

Luminita RADULESCU, Cristian MARTU, Tudor RADULESCU, Lucia Corina DIMA-COZMA, Oana BITERE, Corina BUTNARU, Horatiu STEFANESCU, Sebastian COZMA

Revista de cercetare și intervenție socială, 2018, vol. 60, pp. 180-187

The online version of this article can be found at:
www.rcis.ro, *www.doaj.org* and *www.scopus.com*

Published by:
Expert Projects Publishing House

expert projects
publishing

On behalf of:
„Alexandru Ioan Cuza" University,
Department of Sociology and Social Work
and
Holt Romania Foundation
REVISTA DE CERCETARE SI INTERVENTIE SOCIALA
is indexed by ISI Thomson Reuters - Social Sciences Citation Index
(Sociology and Social Work Domains)

Genetic Screening of Deaf Children: Ethical Considerations

Luminita RADULESCU[1], Cristian MARTU[2], Tudor RADULESCU[3], Lucia Corina DIMA-COZMA[4], Oana BITERE[5], Corina BUTNARU[6], Horatiu STEFANESCU[7], Sebastian COZMA[8]

Abstract

Speech development was one of the elements which determined the evolution of the human being as an individual and as a member of a society. Normal hearing at birth is a prerequisite to acquired speech. The paper discusses the ethical problems linked to the use of genetic screening in the diagnosis of hearing loss. To forbid or determine (at request) the birth of a deaf child, to consider deafness to be a culture or a disability, to require the informed agreement for genetic screening are the ethical dilemmas approached in this paper; it is also an analysis of these facts from the point of view of the deaf community and of the people with a normal sense of hearing. The present paper would like to open a debate concerning the recent opportunities which have made possible the early genetic diagnosis and the treatment of deafness with the cochlear implant, the use of cochlear implant leading to the eradication of deafness (as a disability) and therefore, to the disappearance of the deaf culture.

Keywords: ethics, informed consent, hearing loss, genetic, screening.

[1] University of Medicine and Pharmacy "Grigore T. Popa", Faculty of Medicine, Otorhinolaryngology Department, 16 Universitatii Street, 700115, Iasi, ROMANIA. E-mail: lmradulescu@yahoo.com

[2] University of Medicine and Pharmacy "Grigore T. Popa", Faculty of Medicine, Otorhinolaryngology Department, 16 Universitatii Street, 700115, Iasi, ROMANIA. E-mail: cristimartu@gmail.com

[3] University "Al. I. Cuza", Faculty of Law, 11, Bulevardul Carol I, 700506, Iaşi, ROMANIA. E-mail: avocat_tudorradulescu@yahoo.ro

[4] University of Medicine and Pharmacy "Grigore T. Popa", Faculty of Medicine, I[st] Medical Department, 16 Universitatii Street, 700115, Iasi, ROMANIA. E-mail: cdimacozma@yahoo.com (Corresponding author)

[5] Rehabilitation Clinical Hospital, Audiology Department, 14, Pantelimon Halipa, Iasi, ROMANIA. E-mail: oana.bitere@gmail.com

[6] University of Medicine and Pharmacy "Grigore T. Popa", Faculty of Medicine, Otorhinolaryngology Department, 16 Universitatii Street, 700115, Iasi, ROMANIA. E-mail: cmbutnaru@yahoo.com

[7] University of Medicine and Pharmacy "Victor Babeş", Faculty of Medicine, Otorhinolaryngology Department, 2 Piaţa Eftimie Murgu, Timişoara, 300041, ROMANIA. E-mail: hstefanescu@yahoo.com

[8] University of Medicine and Pharmacy "Grigore T. Popa", Faculty of Medicine, Otorhinolaryngology Department, 16 Universitatii Street, 700115, Iasi, ROMANIA. E-mail: scozma2005@yahoo.com

Introduction

Normal hearing at birth is one of the prerequisite conditions for speech acquisition. In this context, early identification and treatment of deafness (the most frequent sensorial pathology encountered at birth – 1-3‰), represents one of the most important preoccupations for the public health policy in the developed countries. Progress in medical technology has created new opportunities in the treatment of deafness, allowing us to promptly intervention. Thus there are three main medical conduct pathways. The first is early identification of children with hearing loss using electrophysiological methods (otoacoustic emissions – OAE, brainstem evoked response audiometry – BERA, auditory steady state response – ASSR). Such methods lead to the diagnosis of congenital deafness from the first days after birth (Johnson *et al.*, 2005). They are non-invasive, relatively easy to perform and are highly available for usage as screening tools for deafness in maternities (Norton *et al.*, 2000).

The second pathway takes into account the new possibilities of deafness treatment either with powerful conventional digital hearing aids or with implantable prosthesis for the middle ear (like BAHA - bone anchored hearing aid), for the internal ear (cochlear implant) or for the eighth nerve (brainstem implants) (Davis, 1997; Radulescu & Martu, 2007).

New and improved standards of treatments in different diseases have become available as technology progresses. Thus DNA decoding created the possibility for prenatal diagnosis in different diseases like deafness by identifying the gene mutations responsible for hearing impairment, such as partial or total loss of hearing (Abe, Yamaguchi & Usami, 2007; Hone & Smith, 2003; Schade *et al.*, 2003; Sugata *et al.*, 2002; Toader, 2010). Genetic mutations are encountered in 60% of congenital deafness. Molecular diagnosis of deafness in families with hearing impaired children could predict the probability of a mutation able to be transmitted to other members of the patient's family, being the reference point for genetic counseling and, eventually, for prenatal diagnosis of deafness, at the parent's request (Coviello *et al.*, 2004; Marpeau, 2008). This approach opens a real possibility for deafness eradication. The third pathway consists of molecular diagnosis used to establish the etiology of deafness in some countries.

The first pathways described are already implemented in our country. The third pathway consisting of molecular screening in children with hearing loss followed up by genetic counseling and eventual prenatal molecular diagnosis in siblings is pending. Genetic mechanisms also seem to be involved in the pathogenesis of deafness caused by middle ear inflammatory diseases. Pediatric cholesteatomas are usually more aggressive and invasive; as demonstrating in studies genes important in inflammatory processes (for example, KRT6B, SPP1 and S100A7A) are highly up regulated in cholesteatoma (Maniu *et al.*, 2014). The A1555G mutation in

the mitochondrial RNA gene has been associated with aminoglycoside induced hearing loss (Moroti *et al.,* 2009).

Ethical challenge in the genetic management of deafness

In the context of molecular screening of hearing loss some questions arise: (1) Is it ethical to forbid the birth of a deaf child? (2) Can we integrate deafness into a culture or should we consider it to be a disability? (3) Is it ethical to use genetic information regarding the birth of a child according to his/her hearing condition? (4) It is well known that there are deaf people who have the desire to give birth to a deaf child? (5) Is the cultural identity of deaf people in danger of disappearing? (6) Is it necessary to have the informed consent for molecular diagnosis of deafness?

Variability is a prerequisite of evolution. This law of biology leads to the evolution of species by natural selection. To be different from normal can offer unexpected and unknown advantages to a human being or to a group of people provided the difference is not a disadvantage. Evolution regarding human life in modern society with developed medicine is an abstract concept.

Further on, we may ask ourselves what direction the human species will follow – giving way to diversity including evolution without interfering with the human genome or having it under control and guiding it towards perfection. Having diversity in view, one can ask if deafness has any advantages. It is known, for example, that sickle cell anemia provides some resistance to malaria. It is also a fact that people with Down syndrome are protected from some forms of cancer (Hasle, Clemmensen & Mikkelsen, 2000).

Are there any advantages in being deaf? Is it ethical to give birth to people with this disability (or to do nothing in order to prevent their conception) just for the sake of a natural experiment? The immediate benefit and good health should be the moral guidelines. Can we integrate deafness into a culture or should we consider it to be a disability? Authors like Johnson T. (Johnson, 2004; Johnston & Schembri, 2007) supported the view that deafness is not a disability. Those who support the deaf culture say that deafness unlike other disabilities has its own language – sign language – therefore creating, in turn, a linguistic community. Having this in view, deafness is not a disability (Johnson, 2004; Johnston & Schembri, 2007; Padden & Humphries, 1988).

As a result, the pathological concept is inadequate and the only way to understand the status of deafness is to be deaf yourself. In a study from 2001 conducted by the Study Center of Deafness in Bristol it is shown that the majority of deaf persons consider deafness as a disability (Dye *et al.*, 2001). From the point of view of a deaf child it is not fair that other children have a sense they do not have. In such a context persons who can hear are cheaters. If we could choose between being deaf and having the sense of hearing, what would be our option? The advantage of being

able to hear or the lack of this capability? The study carried out in Bristol (Dye *et al.*, 2001) certifies that this conclusion is valid for a large number of deaf people. Asking a deaf person, "would you rather hear?" has no sense and could not have any answer. It is not possible to limit deafness to the absence of only one sense. A deaf person cannot imagine being something he has never experienced. The life of a deaf person may be empty or like the one of any other person who can hear.

For a child with deaf parents – this disability is virtually non existent in the first part of his/her life and in the beginning life experience can be normal. Later on, communication with persons with normal hearing, hence with the whole society, becomes a problem and deafness creates limitations and difficulties as far as life opportunities are concerned. Is it ethical to intend to have or to determine the birth of a child who will be deaf?

If deafness is considered not to be a disability, then there are no ethical problems for the person who wants to have a deaf child. But, as the majority of people consider deafness to be a disability, an ethical dilemma might arise. In Nazi Germany deaf persons were not allowed to get married, such persons were forced to be sterilized or were even killed (Schuchman, 2004).

Advantages and limitations of genetic screening for deafness

Today the situation is totally different, hearing loss screening and early treatment make it possible for a person to be able to hear. Bioethics studies evaluated the moral values and concepts to be included in decision making. In a complex moral universe, a moral code should secure maximum personal happiness for everyone. In the universal moral code a minimum standard should include the principles of not harming and of being good (the principle of beneficence). According to the highest principle of autonomy the patient is in the most favorable position to obtain all that can satisfy him and make him happy as an individual. This principle cannot be applied to a young child because his parents are the ones who should decide for him. It is to be understood that each parent wants what is the best for his child. Therefore, a parent cannot desire (according to the principle of beneficence for his child) to give birth to a child with a predictable but avoidable disability.

However, if the parents do not perceive deafness as a disability, then their desire to have deaf children is to be understood (Murray, 2004). If we accept that deafness is not a disability the position held by such parents can be ethical. On the other hand, it is not necessary to create deaf children just to perpetuate the culture and language of the deaf people.

In countries with advanced medicine, the deaf community is shrinking and therefore the desire to have deaf children to preserve the community is increasing. Nevertheless, to secure a culture based on a disability cannot be justified. Do parents have the right to select their children based on their hearing status? The

human biological right implies five intact and functional senses at birth. With this requirement, the conception of a child with limited sensorial capacities is a violation of this human biological right. We do not have the right to choose deafness, taking into account the future consequences for the child.

The right to have an open future, for as long as possible (an open future means not to limit or confine in any way the life endowment) does not apply to a child who is born with a disability. A deaf child with deaf parents can discover in his past the following: (1) parents knew that their child might be deaf but did not take either the hearing loss or the genetic screening and/or reproductive screening for diagnose and early treatment of deafness; (2) parents took the hearing loss screening but did nothing to rehabilitate their child; (3) parents used genetic techniques to be sure that their child will be deaf. The deaf child can accept the situation, or to claim sanctions or compensations from those he/she considers to be responsible for his/her condition (parents, doctors), for his/her suffering and for his/her limited chances. This scenario is more and more possible.

Is it ethical to use genetic tests and reproductive techniques to decide the birth of a child according to his auditory status? The knowledge and techniques in the field of genetics have made rapid progress and the information provided by mass media has promoted the opportunities in this domain (Toader, 2010). Brugner (Brugner, Murray, O'Riordan, Mathews, Smith & Robin, 2000) in a study conducted in the USA in 2000, shows that 87% persons are willing to have genetic prenatal tests; Martinez in a study from 2003 indicates a percentage of 64% (Martinez, Linden, Schimmenti & Palmer, 2003), Middleton indicates that 28% of the deaf people are willing to have prenatal genetic tests (Middleton, 2004). The aim of genetic and reproductive techniques is to create an embryo without the genes of deafness or to detect the presence of such genes in the embryo.

In many cultures, societies or religions the use of such techniques is justified from a medical and ethical point of view. In some other areas, abortion is not allowed; in such cases for couples at risk, a solution could be the selection of germinal cells to create an embryo without disabilities. In this way, for those who do not accept abortion, the scenario implying avoiding deafness by genetic screening and reproductive techniques responds to the most important ethical criteria. Religion is a major component of communities and was involved in providing recommendations on various modern treatments, maintaining a preferable dialogue between doctors and patients (Dima-Cozma & Cozma, 2012).

From the legal point of view, the molecular screening of hearing loss may be based on Article 34, 1st paragraph of the Constitution, which states that the "right to defend health is guaranteed" and in the 2nd paragraph where it is stipulated that "the state is obliged to take measures in order to ensure the population's hygiene and health". The molecular diagnosis in a child is possible only after having the agreement of the legal tutor and only if the screening is done in the benefit of the

child or if the result of the test identifies the predisposition to a certain disease for the tested child or for his future siblings (Stafie, 2009).

This fact may be beneficial for health if preventive precautions can be taken or the risks can be diminished by modifying the surrounding environmental factors, the style of life or the person's behavior or if there is the possibility to apply an early treatment. Similar to other categories of chronic illness, some stressors can act during the gestational period, determining uterine growth delay and associated malformations. Social factors and quality of life could be related with the continuation and escalation of chronic diseases, in general and with deafness in particular (Dima-Cozma, Mitu, Szalontay & Cojocaru, 2014).

In the Additional Protocol of the Convention for Human Rights and Dignity of the Human Being, the genetic tests done for the sake of research in the field of biology and medicine are ruled out.

Conclusion

In conclusion, genetic screening of deafness should be done in accordance with the objectives of the National Programme. Its aims coincide with those of "The Principles of the Patient's Rights in Europe", which have been internalized through the adoption of the Law number 46 from 2003 regarding the patient's rights, among which, it is recognized the right "for medical assistance of the highest quality".

Acknowledgements

This study was supported by the grant ASM-ANCSI-2016, contract no. 25BM/2016. All authors contributed equally to this paper.

References

Abe, S., Yamaguchi, T. & Usami, S. (2007). *Genetic Testing*, *11*(3), 333-340.

Brugner, J.W., Murray, G.S., O'Riordan, M., Mathews, A.L., Smith, R.J.H. & Robin N.H. (2000). Parental attitudes toward genetic testing for pediatric deafness. *American Journal of Human Genetics*, *67*, 1621-1625.

Coviello, D.A., Brambati, B., Tului, L., Percesepe, A., Sironi, F., Sahai, A., … & Forabosco, A. (2004). First-trimester prenatal screening for the common 35delG GJB2 mutation causing prelingual deafness. *Prenatal Diagnosis, 24*(8), 631-634.

Davis, D.S. (1997). Cochlear implants and the claims of culture? A response to Lane and Grodin. *Kennedy Institute of Ethics Journal*, *73*, 253-258.

Dima-Cozma, C., & Cozma, S. (2012). Religion and medicine or the spiritual dimension of healing. *Journal for the study of religions and ideologies*, *11*(31), 31-48.

Dima-Cozma, C., Mitu, F., Szalontay, A., & Cojocaru, D.C. (2014). Socioeconomic status and psychological factors in patients with essential hypertension. *Revista de cercetare şi intervenţie socia*la, *44*, 147-159.

Dye, M., Kyle, J.G., Allsop, L., Dury, A. & Richter, J. (2001). Deaf people in the community: *Health and disability*, Bristol, U.K.: Deaf Studies Trust.

Hasle, H., Clemmensen, I.H. & Mikkelsen, M. (2000). Occurrence of cancer in individuals with Down syndrome. *Ugeskr Laeger*, *162*(34), 4535-4539.

Hone, S.W. & Smith, R.J. (2003). Genetic screening for hearing loss. *Clinical otolaryngology and allied sciences, 28*(4), 285-290.

Johnson, J.L., White, K.R., Widen, J.E., Gravel, J.S., James, M., Kennalley, T., … & Holstrum, J. (2005). A multicenter evaluation of how many infants with permanent hearing loss pass a two-stage otoacoustic emissions/automated auditory brainstem response newborn hearing screening protocol. *Pediatrics, 116*(3), 663-672.

Johnston, T. & Schembri, A. (2007). *Australian Sign Language (Auslan): An introduction to sign language linguistics*. Cambridge: Cambridge University Press.

Johnston, T. (2004). W(h)ither the deaf community? Population, genetics, and the future of Australian sign language. *American Annals of the Deaf*, *148* (5), 358-375.

Maniu, A., Harabagiu, O., Perde Schrepler, M., Catana A., Fanuţa, B. & Mogoanta C.A. (2014). Molecular biology of cholesteatoma. *Romanian Journal of Morphology and Embryology*, *55*(1), 3-6.

Marpeau, L. (2008). About precocious prenatal screening for profound deafness. Gynécologie, *obstétrique, fertilité & sénologie*, *36*(7-8), 711.

Martinez, A., Linden, J., Schimmenti, L.A. & Palmer, C.G.S. (2003). Attitudes of the broader hearing, deaf and hard of hearing community toward genetic testing for deafness. *Genetics in Medicine*, *5*, 106–112.

Middleton, A. (2004). Deaf and hearing adult's attitudes toward genetic testing for deafness. *Genetics, disability, and deafness*. J. van Cleve (ed.), Washington, DC: Gallaudet University, 127–147.

Moroti Constantinescu, V.R., Georgescu, M., Kovacs, E., Hristea, A., Arama, V. & Pascu, A. (2009). Aminoglycoside-induced destruction of the cochlea. *Therapeutics, Pharmacology and Clinical Toxicology*, *XIII* (2), 210-214.

Murray, J.J. (2004). True love and sympathy: The deaf-deaf marriages debate in transatlantic perspective. *Genetics, disability, and deafness*. J. van Cleve (Ed.) Washington, DC: Gallaudet University, 42–71.

Norton, S.J., Gorga, M.P., Widen, J.E., Folsom, R.C., Sininger ,Y., Cone-Wesson, B. … & Fletcher, K. (2000). Identification of neonatal hearing impairment: evaluation of transient evoked otoacoustic emission, distortion product otoacoustic emission, and auditory brain stem response test performance. *Ear and hearing*, *21*(5), 508-528.

Padden, C.A. & Humphries, T. (1988). *Deaf in America: Voices from a culture*. Cambridge, MA: Harvard University Press.

Radulescu, L. & Martu D. (2007). Do we need an ethics committee in order to make decisions regarding the cochlear implant? *Revista Romana de Bioetica, 5*(2), 27-32.

Schade, G., Kothe, C., Ruge, G., Hess, M. & Meyer, C.G. (2003). Non-invasive screening for GJB2 mutations in buccal smears for the diagnosis of inherited hearing impairment. *Laryngorhinootologie*, *82*(6), 397-401.

Schuchman, J.S. (2004). Deafness and eugenics in the Nazi era. In J. van Cleve (Ed.), *Genetics, disability, and deafness.* Washington, DC: Gallaudet University, 72–78.

Stafie, C. (2009). Therapeutic patient education for the self-management of chronic diseases. *Revista Romana de Bioetica, 7*(2), 103-107.

Sugata, A., Fukushima, K., Sugata, K., Fukuda, S., Kimura, N., Gunduz, … & Nishizaki, K. (2002). High-throughput screening for GJB2 mutations--its clinical application to genetic testing in prelingual deafness screening for GJB2 mutations. *Auris Nasus Larynx*, *29*(3), 231-239.

Toader, E. (2010). Ethics in medical technology education. *Revista Roman*a *de Bioetic*a, *8*(2), 157-162.