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COCHLEAR IMPLANTS AND PSYCHIATRIC ASSESSMENTS: A NORRIE DISEASE CASE REPORT

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SUMMARY

Background: *It is important to perform psychiatric assessments of adult patients who are candidates for cochlear implants both to screen them for psychiatric disorders and to assess their understanding and compliance with the procedure. Deafness is a factor of difficulty for conducting in-depth psychiatric interviews, but concomitant blindness may make it impossible.*

Subjects and method: *After a description of Norrie disease, a rare disease in which blindness and deafness may occur together, we propose a case report of a patient suffering from the disease and who consulted in view of a cochlear implant.*

Results: *Early information on cochlear implants appears to be necessary before total deafness occurs in patients suffering from Norrie disease. An inventory of digital communication tools that can be used by the patient is also highly valuable.*

Conclusions: *Research should be supported for a more systematic use of psychiatric assessments prior to cochlear implants. In the special case of Norrie disease, we recommend early screening for mental retardation and related psychotic disorders and, depending on the patient's level of understanding, preventive information on the benefits and limits of cochlear implants before total deafness occurs.*

Key words: *cochlear implant – Norrie disease – liaison psychiatry*

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INTRODUCTION

In the context of multidisciplinary work at an audiophonology center, the two most common issues are problems of deafness and tinnitus. We have already seen the clinical interest of liaison psychiatry in the assessment and monitoring of chronic tinnitus (Jacques et al, 2013) and the need for it to be integrated with ENT monitoring.

Concerning deafness arising in adults, an audiophonology center will establish an analysis to assess the causes of deafness and may determine an indication for a cochlear implant.

Given the high cost of the procedure, precise criteria must be met for the cochlear implant operation to be reimbursed.

Furthermore, it is also fundamental for the patient first to fully understand the need for follow-up rehabilitation after the operation when the implant starts to work.

Brain plasticity needs to be stimulated through adaptation and regular learning to use the sounds provided by the implant. Insofar as the patient has developed lip reading as a compensation strategy for deafness, he will, in a way, have to "unlearn" using lip reading alone and adopt the habit of giving priority to sound recognition.

In this context, a systematic in-depth psychiatric assessment is necessary:

- - first, through a search for preexisting psychiatric comorbidities (such as schizophrenia, mood disorders or consumption of problematic substances) or any developed in reaction to deafness (frequently depression and social anxiety).
- -second, an assessment of the patient's understanding of how a cochlear implant works and the need for compliance and sustained commitment to rehabilitation.

It is therefore necessary to be able to establish how certain psychiatric disorders may interfere with understanding and compliance. Beyond proven psychiatric disorders, the question of the patient's ability to understand and the influence his/her emotional state has on such understanding requires sufficient assessment time.

The purpose of our publication is to emphasize the importance of this question, but more importantly to point out the practical difficulties encountered in carrying out this psychiatric assessment.

In some situations, deafness is not total and a discussion with the psychiatrist is possible. The use of written communication (often through the interface of a computer screen) is usually the rule and entails taking more time than for conventional consultations.

Using a specific case report, we will demonstrate how such assessments can become impossible and will then discuss possible preventive measures.

SUBJECTS AND METHODS

The ideas developed in this article are based on a case report of a young, 22-year-old patient suffering from Norrie disease and who consulted the Audiophonology Center at the Centre Hospitalier Universitaire,

UcL Namur, in Godinne, Belgium, to consider having a cochlear implant.

After describing Norrie disease based on a review of the literature on Medline, we will describe the factors that made it impossible to perform a psychiatric assessment of this patient suffering from Norrie disease. We will describe alternatives that could be used and will discuss specific preventive measures that could be envisaged in the context of Norrie disease for using cochlear implants.

RESULTS

Norrie disease, also called Norrie-Wardburg disease, is a rare vitreoretinal pathology connected to X characterized by abnormal retina development with congenital blindness. Typical associated manifestations include sensory hearing loss and developmental delay, mental retardation and/or developmental disorders.

The salient clinical feature early in life is a dense, white, vascularized mass behind each lens due to maldeveloped retina. Cataracts and corneal opacities are developed in young childhood, followed by bulbar atrophies (Ohba et al, 1996).

The disease has variable clinical presentations related to the mutation of the gene involved (Berger et al, 1992) and deficits in vascular development appear to be behind the eyesight and hearing problems (Rehm et al, 2002).

Prevalence and annual incidence are unknown, but more than 400 cases have been described. The patients are almost always men, although women vectors do exist. Visual impairment ranges from perception of light to complete congenital blindness. Most of the men affected develop progressive asymmetrical sensorial deafness that begins during childhood (median age of appearance: 12 years), sometimes early, starting in the first year of life (Skevas et al, 2002). Hearing loss may be severe and bilateral at adulthood. Developmental delay and mental retardation are observed in 20 to 30% of all cases. Some have cognitive disorders or psychosis. (Ohba et al, 1996).

As the main characteristics of Norrie disease have been described, we will now present our case.

A mother came to the audiophonology center with her 22-year-old son suffering from Norrie disease for a consultation concerning his complete deafness. The patient was described as not having any mental retardation nor any behavioral problems. He received good care and support from his family. He had been blind since childhood and had compensated for this by learning Braille, receiving an adapted education and using a computer equipped with translation software enabling him to communicate by e-mail.

His hearing problems began several years ago and he began to withdraw from society as his hearing loss progressed. The patient was dependent on this family, which enabled him to continue functioning with daily support from his loved ones. In the last 6 months, his deafness had reached such a point that interaction was no longer possible, with complete withdrawal from society that worried his mother so much that she decided to bring him to a consultation to assess the possibility of a cochlear implant.

During the first psychiatric consultation, the patient's mother's heteroanamnesis was obviously possible and provided information on his medical history. There was a high degree of family anxiety given the loss of two fundamental channels of communication: hearing and eyesight. Interactions by touch enabled the family to communicate and to deal with everyday life. The patient could talk, but the intensity of his voice was maladjusted.

Given this particular context, it was not possible to perform an individual psychiatric assessment as the usual strategy of using a computer screen to communicate was made impossible by the patient's blindness.

Thanks to the patient's Braille translation software, the assessment was performed remotely by e-mail at two scheduled times and with a final face-to-face consultation using questions sent in advance by e-mail. Without this system, it would have been impossible to determine the patient's level of understanding and acceptance concerning the possibility of receiving a cochlear implant.

DISCUSSION

In the literature, emphasis is placed on the difficulty of diagnosing Norrie disease, given how rare it is. Genetic research on the mutations of the gene involved in Norrie disease (Gal et al, 1988) has now made prenatal diagnosis possible (Wu et al. 2017).

Prevention and prediction have become possible through a better understanding of this rare disease.

In the literature we do not find any analysis of the disease's psychological and psychiatric consequences, also due to its rareness.

Our case report sheds light on the fact that patients and their families first have to mobilize consequential adaptation strategies to deal with blindness.

The psychological question of acceptance and adaptation to a disability can be viewed from this angle. In this particular case, the psychological suffering had become even more intense with the appearance of a second major handicap when the patient reached adulthood.

In the situation described here, the patient did not demonstrate any mental retardation or prior psychiatric disorders, but without the patient's Braille translation software it would have been impossible to assess, inform and prepare the patient well for a cochlear implant.

Our reflections led us to envisage informing patients with Norrie disease and their families earlier on of the

possibilities and limits of using cochlear implants as a preventive measure.

Two major difficulties must be taken into account: the difficulty of talking about preventive measures for a second potential disability when the patient is already dealing with a difficult situation, and the risk of causing iatrogenic anxiety. Hanging in the balance are the possibly consequential risks of operating on a patient whose understanding and consent are impossible to assess.

CONCLUSIONS

Our reflections based on our case report and a review of the literature have led us to support further research for more systematic use of psychiatric assessments for adult patients who are candidates for cochlear implants. Liaison psychiatry at an audiophonology center should develop technological supports to compensate for the deafness developed by the patient so as to be able to hold assessment interviews in written form. Our case report highlights the difficulty of assessing patients who also suffer from blindness.

In the context of Norrie disease, even though this disease is rare, the increased related risk of mental retardation and psychosis leads us to recommend that early information should be provided on the risks of deafness and on the possibilities and limits of cochlear implants while the patient maintains good hearing. Digital communication strategies should also be encouraged.

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All authors make substantial contributions to conception and design, and/or acquisition of data, and/or analysis and interpretation of data.

References

1. *Berger W, van de Pol D, Warburg M, Gal A, Bleeker-Wagemakers L, de Silva H, Meindl A, Meintinger T, Cremers F, Ropers HH: Mutations in th candidate gene for Norrie disease. Hum Mol Genet 1992; 1:461-465.*
2. *Chen ZY, Denney RM, Breakfield XO: Norrie disease and MAO genes: nearest neighbors. Hum Mol Genet 1995; 4: 1729-1737.*
3. *Gal A, Uhlhaas S, Glaser D, Grimm T: Prenatal exclusion of Norrie disease with flanking DNA markers. Am J Med Genet 1988; 31:449-453.*
4. *Jacques D, Nozeret Y, Zdanowicz N, Reynaert C, Garin P, Gilain C: Tinnitus and psychiatric comorbidities in liasion psychiatry, analysis of three years in an audiophonology center. Psychiatr Danub 2013: 25:S102-104.*
5. *Ohba N, Isashiki Y: A literature review of Norrie disease. Nippon Ganka Gakkai Zasshi 1996; 100:101-110.*
6. *Rehm HL, Zhangs DS, Brown MC, Burgess B, Halpin C, Berger W, Morton CC, Corey DP, Chen ZY: Vascular defects and sensorineural deafness in a mouse model of Norrie disease. J Neurosci 2002; 22:4286-4292.*
7. *Skevas A, Kastanioudakis I, Daniilidis B, Exarchakos G: Norrie-Wardburg syndrome. Laryngorhinotologie 1992; 71: 534-536.*
8. *Wu LH, Chen LH, Xie H, Xie YJ: Prenatal diagnosis of a case of Norrie disease with late development of bilateral ocular malformation. Fetal Ped Pathol 2017; 10:1-6.*

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