COCHLEAR IMPLANT IN JERVELL AND LANGE-NIELSEN SYNDROME

SUMMARY:
We present a case of cochlear implantation in a child of 3 years and 9 months old with Jervell and Lange-Nielsen syndrome. Jervell and Lange-Nielsen syndrome represents a rare, autosomal recessive cause of congenital deafness. Affected patients have a characteristic prolongation of the QT interval on electrocardiogram, along with cardiac arrhythmias, recurrent syncopal episodes, and a predisposition to sudden death.

Key Words: cochlear implant, Jervell and Lange-Nielsen syndrome, prolongation of the QT interval.

INTRODUCTION

Jervell and Lange-Nielsen syndrome (JLNS) is characterized by congenital profound bilateral sensorineural hearing loss and long QTc, usually greater than 500 msec. Prolongation of the QTc interval is associated with tachyarrhythmias, including ventricular tachycardia, episodes of torsade de pointes ventricular tachycardia, and ventricular fibrillation, which may culminate in syncope or sudden death. The classic presentation of JLNS is a deaf child who experiences syncopal episodes during periods of stress, exercise, or fright. It is known to be associated with mutations of the genes KCNQ1 (KVQTI) and KCNE1 (Isk). (1)

CASE REPORT

We present a case of cochlear implantation in a child of 3 years and 9 months old with Jervell and Lange-Nielsen syndrome. The patient (a girl) presented a profound bilateral sensorineural hearing loss. The audiologically evaluation consisted in pure-tone thresholds – revealed profound bilateral sensorineural hearing loss-, impedance – timpanogram type A-, stapedian reflex – absent-, OAE- absence of OAE-, ABR- absence of wave V-. The cardiology exam revealed extrasystolic ventricular arrhythmia with the prolongation of the QT interval on electrocardiogram. Preoperative evaluation included a temporal bone CT (for evaluation the inner ear, the cochlea was unossified) and infantile neuropsychiatric exam (the development index QD was in normal limits).

We established the clinical diagnostic of Jervell and Lange-Nielsen syndrome based on congenital sensorineural deafness and long QT interval.

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For this profound sensorineural hearing loss we proposed cochlear implantation. It was mandatory that the patient`s parents give their consent. We used classic technique, mastoidectomy and posterior tympanotomy (introduced by W.F. House), cochleostomy and positioning of the electrode array.

Special perioperative precautions were required, as cardiac monitoring. Tolerance to general anesthesia was very good. Perioperative and postoperative evolution was good, without any complication.

Postoperative rehabilitation started 1 month after surgery, with additional evaluations at 3, 6, 9, 12, 18, and 24 months postoperatively and then every year.

This patient has responded well to cochlear implantation. The child has achieved limited open-set word comprehension and significantly improved speech, as is expected for her age.

**DISCUSSIONS**

Jervell and Lange-Nielsen syndrome represents a rare, autosomal recessive cause of congenital deafness. (2) This syndrome seems to affect less than one percent of all deaf children.(3) In the ENT Department of the University of Medicine Timisoara, 66 patients received cochlear implants between January 2003 and January 2010 but only one case was diagnosed with Jervell and Lange-Nielsen syndrome.

Komsouglu B. and all. submitted that fifty percent of individuals with JLNS had cardiac events before age three years.(4)

Schwartz and all. claimed that females are at lower risk for cardiac arrest and sudden death. Most mutations (90.5%) are on the KCNQ1 gene; mutations on the KCNE1 gene are associated with a more benign course. Subgroups at relatively lower risk for cardiac arrest and sudden death include females, patients with a QTc < or = 550 ms, those without events in the first year of life, and those with mutations on KCNE1.(5)

Cochlear implantation can be relatively safely performed in patients with Jervell and Lange-Nielsen syndrome, provided proper precautions are followed.(2)

**CONCLUSION**

The clinical diagnostic of Jervell and Lange-Nielsen syndrome is based on congenital sensorineural deafness and long QT interval.

The genetic diagnostic, if is possible, included three categories of indication: genetic diagnostic in course of differential diagnostic, predictive genetic diagnostic and prenatal genetic diagnostic. It is mandatory that the patient give his full consent.(6)

Perioperative and postoperative evolution was good, without any complication. This could be explain by the presence of the positive predictive factors (the patient was female and she didn`t have cardiac events in her history).

This patient has responded well to cochlear implantation. The child has achieved limited open-set word comprehension and significantly improved speech, as is expected for her age.

Cochlear implantation can be relatively safely performed in patients with Jervell and Lange-Nielsen syndrome and the children received significant benefit from cochlear implantation.

**References:**