International Journal of Pediatric Otorhinolaryngology 73 (2009) 889–894

Contents lists available at ScienceDirect

International Journal of Pediatric Otorhinolaryngology

journal homepage: www.elsevier.com/locate/ijporl

Case report

Bilateral cochlear implantation in children with Noonan syndrome

C. Scheiber [a,*], A. Hirschfelder [a], S. Gräbel [a], H. Peters [b], H. Olze [a]

[a] Klinik für Hals-, Nasen- und Ohrenheilkunde, Charité – Universitätsmedizin Berlin, Campus Virchow-Klinikum, Augustenburger Platz 1, 13353 Berlin, Germany
[b] Institut für Medizinische Genetik, Charité – Universitätsmedizin Berlin, Germany

ARTICLE INFO

Article history:
Received 14 October 2008
Received in revised form 16 February 2009
Accepted 18 February 2009
Available online 19 March 2009

Keywords:
Noonan syndrome
Cochlear implant
Hearing disorder
Deafness
Inherited disorder

ABSTRACT

Noonan syndrome is a mostly autosomal dominant inherited disorder, which can be accompanied by hearing disorders or deafness, coagulation disorders, combined heart defects and developmental disorders. We are reporting on two children with an established Noonan syndrome with a severe bilateral hearing loss of respectively 95 and 100 dB and proper findings in the CT/MRI of the petrous bone. After complete otologic and radiologic diagnostics, both children underwent bilateral cochlear implantation successfully. According to the authors' knowledge, this is the first time that cochlear implant therapy is discussed in patients with Noonan syndrome.

1. Introduction

ENT specialists are frequently confronted with hearing disorders in the context of syndromes. Noonan syndrome is one of these syndromes that has increasingly moved into focus due to new diagnostic developments. It was described for the first time in 1963 as an independent syndrome by Jacqueline A. Noonan, a pediatric cardiologist at the University of Iowa and Dorothy Emke, pediatrician, in a series of patients with characteristic facies and multiple abnormalities [1]. The syndrome is also known under the names of XX-Turner-phenotype, XY-Turner-phenotype and further synonymously used names (Table 1), as it is a complex familial abnormalities syndrome, which clinically resembles the Ullrich–Turner-syndrome in multiple characteristics and appears in both genders [2]. The syndrome's symptoms can include short stature, facial dysmorphias and hearing disorders or even deafness (Table 2). Confirmation of possible hearing disorders and facial dysmorphias lead to consult an ENT specialist. The literature concerning epidemiologic data on the frequency of hearing disorders is rare and to the authors' knowledge there is no comprehensive published data regarding the hearing evolution in patients with Noonan syndrome. Qui et al. [3] examined 20 patients (12 male, 8 female, age 5–65 years, mean 18.6 years) with Noonan syndrome and diagnosed a hearing loss for high frequencies in 50% and for low frequencies in 20% of the patients. In connection with tympanometry and bone conduction threshold, the hearing loss was sensorineural in 75%. The results of pure tone audiometry were widely spread, ranging from normal hearing to severe hearing loss. In a clinical study on Noonan syndrome, Sharland et al. [4] found hearing disorders in 40% of the 151 patients (83 male, 68 female, mean age 12.6 years). The levels of hearing loss were not specified, however. In comparison, numerous abnormalities of the petrous bone in Noonan syndrome have been described in the literature [5–7] (Table 1). According to the authors' knowledge, this is the first time that the aspects of diagnostics and therapy are discussed and the literature is generated regarding cochlear implantation in patients with Noonan syndrome. In the case report, we present the successful bilateral implantation of a Cochlear Implant (CI) in two children with Noonan syndrome.

2. Case report

2.1. Clinical history and clinical findings

We are reporting on two girls (2 years and 4 years) who have been diagnosed with Noonan syndrome at the Charité University Children's Hospital in 2007 on the basis of a cytogenetic examination and the characteristic findings constellation. In both children the karyotype was normal with 46, XX and analysis confirmed the presence of mutation in gene PTPN11. Exons 1–15 of the PTPN11 gene and the flanking intronic sequences were amplified by PCR and bidirectionally sequenced. Apart from the

* Corresponding author. Tel.: +49 30 450 555 002; fax: +49 30 450 555 900.
E-mail addresses: christian.scheiber@charite.de, heidi.olze@charite.de (C. Scheiber).

0165-5876/$ – see front matter © 2009 Elsevier Ireland Ltd. All rights reserved.
doi:10.1016/j.ijporl.2009.02.016

Table 1
Synonyms.

Male Turner syndrome
Familial Turner syndrome
Female pseudo-Turner syndrome
Pseudo-Turner syndrome
Pseudo-Ullrich–Turner-syndrome
Turner-like syndrome
Turner's phenotype with normal karotype
Turner's syndrome in female with X-chromosome
Ullrich–Noonan syndrome
XX Turner phenotype syndrome
Turner phenotype syndrome

appearance which was characteristic for Noonan syndrome, the girls had multiple malformations of the cardio-vascular system (pulmonary valve stenosis, patent ductus arteriosus, atrial septal defect, patent foramen ovale), short stature and a general developmental delay. Weight and height constantly remained considerably under the 3rd percentile and a statomotoric retardation was found.

2.1.1. Case report 1

The 4-year-old girl, a premature infant (36 + 2 weeks of pregnancy [WOP]), was delivered via emergency cesarean section because of a suspected uterine rupture. Postpartum she demonstrated a respiratory adjustment disorder after initial

Table 2
wide clinical variability of symptoms. Symbols are indicating presence (+) and absence (−) of symptoms.

	Case report 1: the 4-year-old girl	Case report 2: the 2-year-old girl
Physical appearance		
Short stature	+	+
Triangular contour of the face	−	+
Short webbed neck with pterygium colli	+	+
Hypertelorism	+	+
Ptosis	+	+
Epicanthus medialis	+	+
High anterior hairline	+	+
Low posterior hairline	+	−
Curly hair	−	+
Mild mental retardation	+	+
Malformations of the cardio-vascular system		
Pulmonary valve stenosis	+	+
Patent ductus arteriosus	+	+
Atrial septal defect	+	+
Patent foramen ovale	+	+
ENT Features		
Hearing disorder	+	+
Low set posteriorly angulated ears	+	+
with thickened helix	+	+
Anatomic middle-	−	−
and inner ear abnormalities	−	−
Petrous bone: absence of the long process of the incus and atypical position of stapes [5]	−	−
Dehiscence of a high jugular bulb, semicircular canal aplasia, extremely large mastoid emissary vein, small and dysplastic cochlea [6]	−	−
Dehiscence of the facial canal, enlarged semicircular canal [7]	−	−
Facial features: High nasal bridge	+	+
Wide nasal base	+	+
Deep philtrum	+	+
Higharched palate	−	−
Micrognathia	−	−
Malalignment of the teeth	−	−
Further symptoms		
Strabismus	−	−
Amblyopia	−	−
Refractive errors	−	−
Cone shaped cornea	−	−
Funnel chest	+	+
Scoliosis	+	+
Simian crease	+	−
Pulmonary valve stenosis	+	+
Testicular maldescent		
Testicular aplasia		
Cryptorchidism		
Seizure disorder	−	−
Thrombocytopaenia and	+	+
Partial deficiencies of coagulation factors XI, XII, XIII	−	−
von Willebrand disease with lack of von Willebrand factor (vWF) and factor VIII	+	−
Individual symptoms		
Leucopenia	−	+
Clinodactyly	+	−
Hypoplastic appearing outer genitals	+	−
A plane occiput	−	+