



## Case Report

# Cochlear Implantation in a Patient with Kabuki Syndrome

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Criteria for cochlear implants are expanding and now include children with disabilities in addition to hearing loss, such as those with Kabuki syndrome (KS). This case report describes language outcomes and changes in the quality of life of a female child with KS after cochlear implantation. The subject had a profound progressive sensorineural hearing loss, cognitive impairments, and other disabilities and communicated using vocalized sounds and the Dutch Sign Language. After cochlear implantation at an age of nine years and three months, the patient displayed no progress in speech production and minimal progress in receptive language development, but she had an increased awareness of the world and an increase in the quality of life.

**KEYWORDS:** Kabuki syndrome, cochlear implantation, intellectual disability, hearing loss

## INTRODUCTION

Hearing impairment is one of the characteristics of Kabuki syndrome (KS) and is seen in up to 50% of the cases <sup>[1]</sup>. For patients with KS and a profound sensorineural hearing loss, a cochlear implant (CI) can be considered. CI placement, however, can be challenging because of the additional handicaps and anomalies of the petrosal bone. To the best of our knowledge, there is no report describing the application of CI in individuals with KS.

One of the aims of this report was to provide an overview of the challenges health professionals may face when considering cochlear implantation in children with KS. The second aim was to investigate whether these challenges are similar to those in other patients with a profound hearing loss in addition to another disability.

## CASE PRESENTATION

The female subject was born in the Netherlands in 2001. We obtained written informed consent from the parents. Her mother was Cuban (Spanish speaking), and her father was Italian (Italian speaking). The parents communicated in Spanish, and neither fluently spoke Dutch. At the age of one year, brainstem-evoked response audiometry indicated auditory thresholds of 55 dB for the right ear and 80 dB for the left ear. In the same year, the subject was diagnosed with KS by a pediatric neurologist (Table 1). From the age of one year and six months to seven years, the subject predominantly wore bilateral hearing aids only at the day care. At the age of eight years, she had progressive hearing loss (Figure 1), and no signs of speech perception were observed.

Computed tomography (CT) showed bilateral enlarged vestibular aqueducts. The right modiolus was incomplete, and the cochlea was classified as incomplete partition type II <sup>[2]</sup> (Figure 2). There was also evidence of dysplasia of the vestibulum and semicircular canals. On the subject's left side, there was evidence of mild dysplasia of the semicircular canals. These abnormalities were confirmed using MRI (Figure 3). MRI also showed that there were no anomalies of the facial or vestibulocochlear nerves.

At the age of six years, the subject began attending a boarding school for deaf children with developmental disabilities, and at the school, she started learning the Dutch Sign Language. At the age of six years and four months a Dutch version of the non-speech test <sup>[3]</sup>, supported by sign language, indicated language development levels corresponding to an 18–21-month-old hearing child. She had an estimated non-verbal IQ of 54 (the Snijders–Oomen nonverbal intelligence test; SON-R2,5-7 <sup>[4]</sup>).

At the age of nine years and six months, the subject was implanted with a CI. The right side was selected because of its better pre-operative hearing abilities and the continued use of amplification compared to those in the left ear. The parents received extensive counseling that the outcome of CI may be low because of the subject's relatively high age for CI, her limited preoperative language skills, and her intellectual disabilities.

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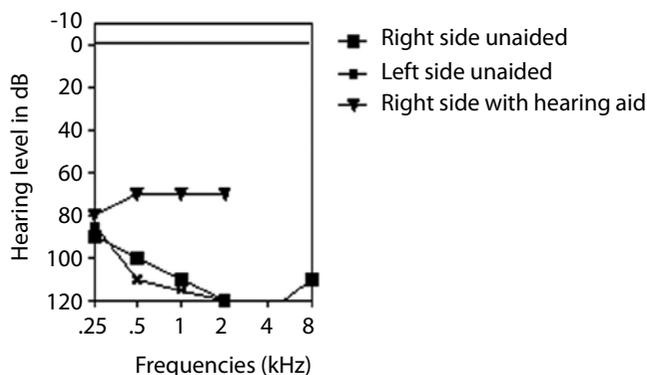


Figure 1. Pure tone audiometry before implantation age: 8 years 7 months

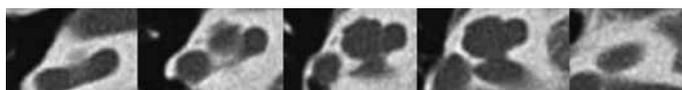


Figure 2. CT of the cochlea right side, incomplete partition type II

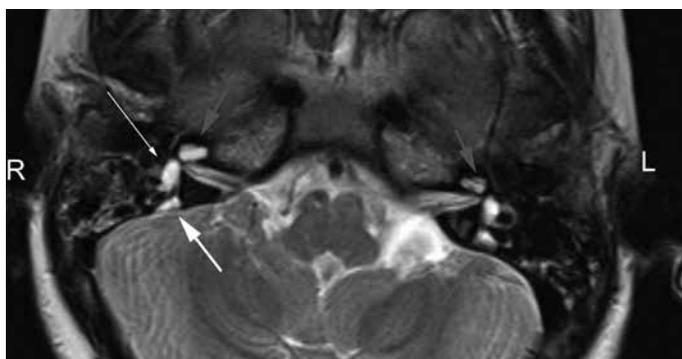


Figure 3. MRI T2; Grey arrows: cochlea; Small white arrow: relatively large vestibulum with dysplasia of the semicircular canals; Big white arrow: enlarged vestibular aqueduct

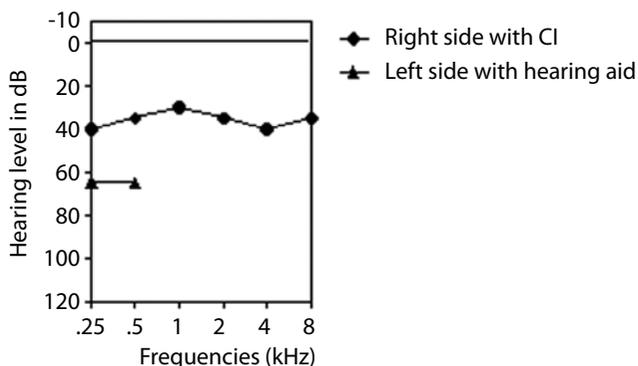


Figure 4. Pure tone audiometry after implantation age: 10 years 9 months. No reaction with the hearing aid on the left side on frequencies of >0.5 kHz

The surgical procedure was performed via cortical mastoidectomy and posterior tympanotomy. During cochleostomy, the perilymph was released under slight pressure, but no real gusher occurred. The cochleostomy was effectively closed with fascia and fibrin glue. A Nucleus 22-electrode CI (Cochlear™ Nucleus® CI512 with contour advance electrode) was completely inserted. Intraoperative stapes reflexes and neural response telemetry of all electrodes showed normal device functioning.

Table 1. A list of the subject's clinical features

Subject's characteristics

- Peculiar facial expression ("Myopathic face")
- Thin upper lip
- High-arched palate
- Uvula bifida
- Blue sclerae
- Left-sided strabismus convergens and a Brown anomaly of the right eye
- General muscular hypotonia
- Hypermobile joints in the extremities
- Short fifth digit on the left hand
- Microcephaly (-3 SD)
- Left-sided congenital hip dysplasia with hip luxation
- Cardiovascular anomalies: a slight pulmonary subvalvular stenosis, multiple septal defects, and an atrial septal defect
- Delayed physical growth and neurodevelopmental skills\*

\*Sit up straight without aid at 13 months, walk at 2 years  
SD: standard deviation

One year after implantation, at the age of ten years and six months, aided auditory thresholds (using headphones) were between 30 and 40 dB (Figure 4). Language comprehension was tested using the Reynell Developmental Language Scales (RDLS) [5]. The subject performed at a developmental level of an 18 month-old. In the test, she could correctly point to some objects on verbal request, but she needed additional sign language to understand more than single-word tasks. In terms of speech production, at this age, she only produced isolated sounds.

At two years six months postoperatively (age of twelve years), the subject's level of RDLS had improved to an age equivalent of 23 months. When language was supported with sign language, the subject's language was at a level comparable to a 3-year-old hearing child. Perceptive speech and language skills continued to slowly progress over this time. At this stage, the subject was receiving frequent training with a speech therapist.

DISCUSSION

The criteria for cochlear implantation continue to expand, and children with plural disabilities are now eligible for CI. Although there are limited studies evaluating the use of CI on communication in children with plural disabilities, the results to date have been promising. We present this case study of a child with KS who had a profound hearing loss in addition to other impairments. The aim of our study was to contribute to the knowledge about the challenges of CI in patients with dual disabilities.

The subject was considered as a candidate for CI because of her level of aided residual hearing in the right ear and her consistent use of a hearing aid throughout her childhood. The expectations on the success of implantation to develop language skills were low given the petrosal bone anomalies, age at implantation, preoperative language abilities, intellectual disability, and the use of more than one language at home.

The surgical procedure is challenging when the inner ear is malformed; however, the malformation observed in our subject was not regarded as a contraindication for cochlear implantation. Despite the incomplete partition of the cochlea, there was sufficient cochlear lumen for the insertion of all electrodes. No intra- or postoperative complications occurred during implantation.

Postoperatively, hearing and speech perception performance skills improved, but receptive and expressive language skills remained below average for the subject's calendar age. The subject required lip reading and sign language to supplement the receptive language. In addition to improved language, the subject had improved recognition of environmental sounds and displayed stronger communicative skills.

Intellectual disability and additional handicaps made it difficult to develop receptive and expressive language skills after implantation<sup>[6]</sup>, besides impairments across the areas of language, speech, and oro-motor functions in patients with KS<sup>[7]</sup>. Regardless of whether a child has a hearing loss only or has a disability in addition, there is a relationship between increased age at implantation and poor performance<sup>[8]</sup>. In this case study, this means that despite the subject's residual hearing during her first years, her age at implantation meant that she had experienced 9 years 6 months with limited aural input.

KS and CHARGE syndrome have a number of similar clinical features<sup>[9]</sup>. The challenges with CI described in our case study for a subject with KS may be applicable to individuals with CHARGE syndrome who are eligible for CI. In other words, both groups have handicaps in addition to hearing loss, and both groups have intellectual disabilities and anomalies of the petrosal bone<sup>[10]</sup>. Given the similarity between the groups and the lack of research on CI in KS, studies investigating CI in individuals with CHARGE syndrome could aid in clinical decision making when it comes to implantation in subjects with KS.

The heterogeneity of KS characteristics ensures that it is difficult to generalize the results of individual cases. Whether a child with KS and a hearing impairment will benefit from CI depends on each individual's profile. Although CI and rehabilitation are challenging, our experience indicates that CI in individuals with KS may assist language development.

**Informed Consent:** Written informed consent was obtained from the parents of the patient who participated in this study.

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