

# Outcomes of Late Implantation in Usher Syndrome Patients

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## Abstract

**Introduction** Usher syndrome (US) is an autosomal recessive disorder characterized by hearing loss and progressive visual impairment. Some deaf Usher syndrome patients learn to communicate using sign language. During adolescence, as they start losing vision, they are usually referred to cochlear implantation as a salvage for their new condition. Is a late implantation beneficial to these children?

**Objective** The objective of this study is to describe the outcomes of US patients who received cochlear implants at a later age.

**Methods** This is a retrospective study of ten patients diagnosed with US1. We collected pure-tone thresholds and speech perception tests from pre and one-year post implant.

**Results** Average age at implantation was 18.9 years (5–49). Aided average thresholds were 103 dB HL and 35 dB HL pre and one-year post implant, respectively. Speech perception was only possible to be measured in four patients preoperatively, who scored 13.3; 26.67; 46% vowels and 56% 4-choice. All patients except one had some kind of communication. Two were bilingual. After one year of using the device, seven patients were able to perform the speech tests (from four-choice to close set sentences) and three patients abandoned the use of the implant.

**Conclusion** We observed that detection of sounds can be achieved with late implantation, but speech recognition is only possible in patients with previous hearing stimulation, since it depends on the development of hearing skills and the maturation of the auditory pathways.

## Keywords

- ▶ usher syndromes
- ▶ cochlear implant
- ▶ delayed diagnosis
- ▶ quality of life

## Introduction

The Usher syndrome is an autosomal recessive disease characterized by varying degrees of sensorineural hearing loss and progressive visual impairment, caused by retinitis pigmentosa (RP).<sup>1</sup>

The syndrome's incidence is estimated as 3–6/100,000 persons in the general population. Among the deaf popu-

lation, the prevalence of the syndrome is 3 to 6%. It is the most frequent cause of adult blindness and deafness. Studies underestimate the true prevalence of the disease due to lack of early diagnosis or for being misdiagnosed. The average age at diagnosis is 10 years and some children are implanted before vision loss and even before the diagnosis of Usher's disease.<sup>1</sup>

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Three distinct subtypes of the syndrome have been described. Usher Type I (USHI) is characterized by profound sensorineural hearing loss at birth or during the first year of life, abnormal vestibular function, and delayed motor development. Children with Usher type II (USHII) have hearing loss from birth moderate to severe and normal vestibular function. Affected children with Usher type III (USHIII) are usually born with normal hearing and undergo progressive hearing loss. Their vestibular function may be affected. The vision loss is gradual and usually begins in adolescence or adulthood.<sup>2</sup> The severity of hearing loss and blindness is higher in Usher syndrome type I.

Several studies of patients with Usher syndrome concluded that cochlear implant (CI) is an effective treatment for such patients, since there is a deterioration of a duplicate sensory. Benefits vary from a better quality of life, observed in questionnaires,<sup>3</sup> to superior performance in oral communication.<sup>2,4-6</sup> This variability of the results is seen as being dependent on factors such as age of implantation, duration of deafness, and type communication prior to implantation.<sup>6,7</sup>

Families have sought the solution for children with Usher syndrome who did not have the opportunity to receive cochlear implants in early childhood, since the visual loss is progressive, starting in adolescence.

We seek to investigate whether late implant patients presenting Usher syndrome type I with pre-lingual hearing loss can benefit the cochlear implant.

**Method**

Among the 1.350 patients implanted in our service, 13 (0.9%) have Usher syndrome, 10 are USHI, and three are USHII. We conducted a retrospective study of data collection. We selected all patients with USHI and excluded patients with USHII plus those who had not been yet implanted. We collected patients' age at the time of surgery, type of communication before and after surgery, hearing thresholds (PTA) pre (with hearing aid) and post implant (1 year of CI use), and the

performance of speech perception pre and post implant. The protocol used to conduct the testing was according to Goffi Gomez et al.<sup>8</sup>

The subjects were classified according to mode of communication. In the case of bilinguals, we set up the first way of communication as the mother language or that of greater fluency.

**Results**

We selected 10 patients implanted (7 men, 3 women) diagnosed with USHI. The average age for implantation was 18.9 years (5–49). ► **Table 1** describes the summary of the characteristics of each patient.

Average pre auditory threshold (PTA HA) was 103 dB HL (► **Fig. 1**). It was possible to measure speech perception in four patients preoperatively (1, 2, 4, and 6), who could only detect speech. Other patients failed to perform the speech perception tests. All patients except one had some kind of effective communication. Two were bilingual (► **Table 2**).

One year after the implantation, the average auditory threshold (PTA CI) was 35 dB (► **Fig. 1**). Only three patients improved their performance in speech perception to 40%, 30%, and 10% for recognition of sentences in closed set, respectively (1, 2 and 3); five patients had only speech detection, and two were not able to carry out tests (► **Table 2**). Three patients abandoned the use of CI (8, 9, and 10).

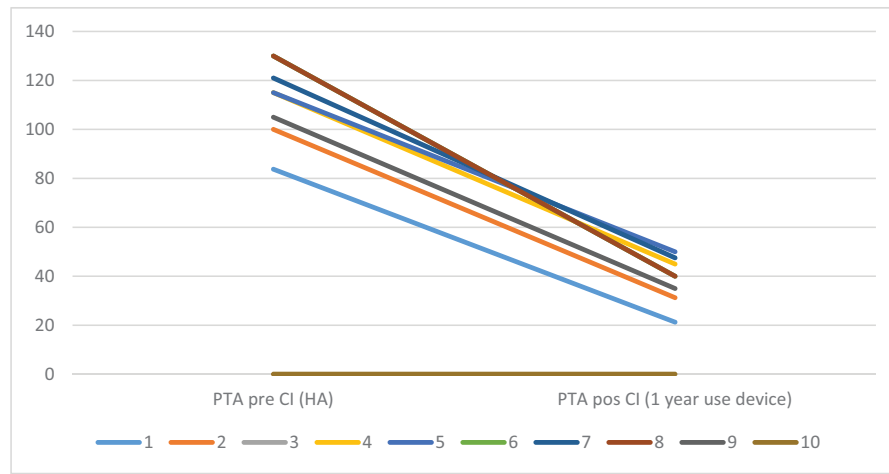
**Discussion**

For proper indication and to establish a prognosis of the cochlear implantation beyond the knowledge of the etiology of deafness patients, it is important to survey the use of hearing aids and residual hearing that allows us to infer the development of the auditory pathway.<sup>9</sup> This will allow us to manage the expectations of the patient and family after cochlear implantation. Several factors can influence the outcome of cochlear implants as age of occurrence / diagnosis

**Table 1** Summary of the characteristics of the 10 patients with implanted Usher syndrome

ID	HA use (years)	Diagnosis age (years)	Implanted age (years)	Side	School	Visual	CI model	Speech strategy
1	11	1	13	R	Mainstream	RP	Cochlear Nucleus 24M/K	ACE
2	7	1	9	R	Mainstream	RP	Cochlear Nucleus 24M/K	ACE
3	10	1	12	R	Mainstream	RP	Cochlear Nucleus Freedom	ACE
4	22	1	23	L	Mainstream	RP	Cochlear Nucleus Freedom	ACE
5	6	2	18	L	Special	RP	Neurelec Digisonic SP	MPIS
6	27	2	29	R	Special	RP	Cochlear Nucleus 24M/K	ACE
7	12	2	49	R	Special	RP	Medel Sonata	CIS
8	1	1	5	L	Special	RP	Cochlear Nucleus 24M/K	ACE
9	5	1	16	L	Special	RP	Cochlear Nucleus 24M/K	ACE
10	13	2	15	L	Special	RP	AB HiRes 90k	HIRESS

Abbreviations: CI, cochlear implants; HA, individual sound amplification device; RP, retinitis pigmentosa.



**Fig. 1** Results of average hearing thresholds pre and post one year of use device.

of hearing loss (pre versus post-lingual) age at implant, residual hearing, systematic use of hearing aids, communication mode, support from family, and educational environment, as well as rehabilitation and personal motivation to join to the world of sound and learn to listen.

Indeed, studies on Usher syndrome reported outcomes of patients implanted with different ages and conclude that patients implanted late have good sound detection thresholds,<sup>2,4,6</sup> but the worst speech perception performance.

The fact that Usher syndrome patients are deprived of another sensory pathway does not necessarily mean that the patients will benefit from this type of rehabilitation. The clinical team should evaluate patients with Usher syndrome carefully and discuss the prognosis both in team and with the family.

The history of the systematic use of hearing aids, residual hearing, and the mode of communication allow us to infer the

value that the patient places on oral communication and listening in their life. They may not develop auditory cortical area and symbolic meaning of sounds. Patients who had no prior central auditory skills because they never had access to sounds when implanted can hardly adapt, leading to treatment failure.<sup>10</sup> The development of oral language skills from birth is essential to the patient subsequently acquire the new language sound of the cochlear implant. They need to use systematically bilateral hearing aids (even with little benefit) to keep the peripheral and central pathways as preserved as possible and learning one or more languages from birth.<sup>11,12</sup> It is possible to the baby with congenital deafness to acquire the natural neurobiological development when the auditory stimulation is restored before 3.5 years of age (Sharma et al., 2009),<sup>13</sup> on the other hand, patients implanted late show changes in the number of projections or synaptic density at various levels of the auditory pathway.<sup>10-14</sup>

**Table 2** Description of communication modes and results of speech perception of 10 patients with Usher syndrome type I

Pre CI				Pos CI (1 year of use device)			
Subjects	Communication mode	PTA HA	Speech perception	Age at CI	Communication mode	PTA CI	Speech perception
1	Sign language/ Oral	83.75	13,3% vowels	13	Sign language/ Oral	21.25	40% closed set
2	Oral/ Sign language	100	26,67% vowels	9	Oral/ Sign language	31.25	30% closed set
3	Sign language	130	NT	12	Sign language	40	10% closed set
4	Oral	115	46% vowels	23	Oral/ Sign language	45	53% vowels
5	Sign language	115	NT	18	Sign language	50	20% vowels
6	Oral	130	56% 4 choice	29	Oral	40	15% vowels
7	Oral	121	NT	49	Oral	47.5	58% 4 choice
8	Sign language	130	NT	5	Sign language	40	NU
9	Sign language	105	NT	16	Sign language	35	NU
10	No language	123.75	NT	15	Sign language	31.25	60% vowels

Abbreviations: CI, cochlear implants; HA, individual sound amplification device; NT, not tested; NU, non user CI patient; PTA CI, pure tone threshold of cochlear implant; PTA HA, pure tone threshold of hearing aid. (no access to speech sounds even with the use of hearing aids).

Detecting the sounds achieved by the majority of our patients show that cochlear implants can provide access to sounds (peripheral input), however, when the implant is delayed, maybe, there are not enough central connections between the auditory area and the association areas. That means that detection is not enough for these people to give meaning to the sounds that they hear and may not be able to achieve the representation of the sounds. Another important factor is the inadequate expectations of the patient and families. Many teens already fluent in sign language also are disappointed with the CI because they might have expected something that had not happened, because the family might have imposed it or that his expectation was beyond that treatment could offer. The cochlear implant teams based on their own experience should judge each case individually to ensure realistic expectations of the outcome. This suggests that traditional measures of speech perception may not be sufficient to accurately reflect the real benefit of CI and alternative assessment tools, including educational, social and psychological areas are needed.<sup>15-19</sup>

## Conclusion

Patients with Usher syndrome who were lately implanted have good hearing thresholds, but speech recognition and use of the device will depend mainly of previous stimulation, since they are directly related to the development of the auditory pathway and the central auditory skills.

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