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EAR SURGERY IN TREACHER COLLINS SYNDROME

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The autosomal dominant hereditary Treacher Collins syndrome manifests itself phenotypically in dysmorphogenesis of particularly the first, but also the second branchial arch system. Consequently, 50% of patients with Treacher Collins syndrome have a congenital, generally pure conductive hearing loss resulting from a major or minor ear anomaly. The outcome of surgery to improve patients' hearing varies and is sometimes even disappointing. Thorough analysis of 33 cases (39 operated ears) and the strict application of a classification for the anomaly to each ear enabled us to gain insight into the most suitable surgical policy and to form a prognosis for reconstructive ear surgery.

KEY WORDS—aural atresia, autosomal dominance, bone-anchored hearing aid, branchial arch, congenital deafness, mandibulofacial dysostosis, ossicular chain anomaly.

INTRODUCTION

Treacher Collins syndrome (TCS), or mandibulofacial dysostosis, is a well-delineated branchial arch syndrome with an autosomal dominant mode of inheritance.[1] However, in about 50% to 60% of cases the family history is negative, and these are thought to be de novo mutations.[2,3]

Treacher Collins syndrome is characterized by antimongoloid slanting of the eyes, coloboma of the lower lid, malar and mandibular hypoplasia, hypoplasia or aplasia of the zygomatic arch, cleft palate, external and middle ear malformations, and conductive deafness. Less common but associated features have also been described, which are summarized in Table 1.

In cases in which all the stigmata are present, it is easy to make a diagnosis on the basis of the clinical appearance (Fig 1). The gene responsible for TCS has only recently been localized on the long arm of chromosome 5.[4]

Conductive hearing loss is thought to be present in about 50% of patients with TCS, and is caused by ossicular chain malformations, often in combination with meatal atresia.[2,5] Mixed or sensorineural hearing loss is rare and has only been reported occasionally.[6-8] Although reconstructive ear surgery can be a challenge for the otologist, the postoperative results are often disappointing.[9]

We present a review of the literature on this subject and additionally describe the otologic findings and surgical management of 12 patients with mandibulofacial dysostosis and conductive hearing loss operated on in the Nijmegen Department of Otorhinolaryngology.

METHODS

We evaluated the otologic data of 12 consecutive TCS patients operated on at the University Hospital Nijmegen between 1960 and 1990 and reviewed the literature on ear surgery in TCS patients published

TABLE 1. FEATURES OF TREACHER COLLINS SYNDROME

Antimongoloid-slanting eyes
Coloboma of lower lid
Malar hypoplasia
Mandibular hypoplasia
Hypoplasia or aplasia of zygomatic arch
Pinna dysplasia
Meatal atresia
Ossicular chain malformations
Cleft palate, high arched palate
Absence of eyelashes on medial part of lower lid
Absence of lower lacrimal puncta
Ear appendages, preauricular sinus
Preauricular hair prolongation
Choanal atresia
Macrostomia
Malocclusion
Obstructive sleep apnea syndrome

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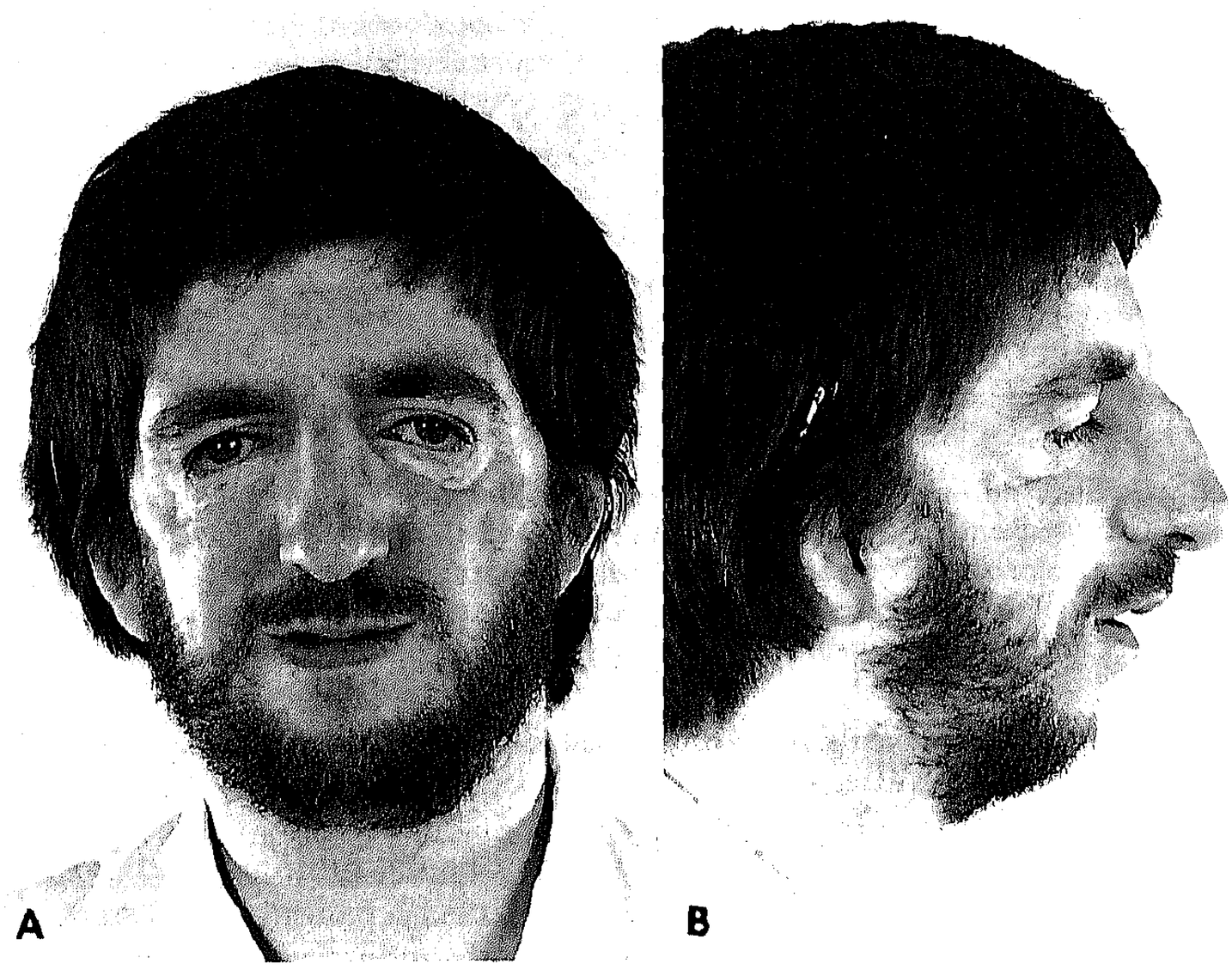

Fig 1. (Case 8) Patient with typical appearance of Treacher Collins syndrome. A) Frontal view. B) Lateral view.

since 1960. This review covered only those reports that provided sufficient details to allow description and classification of the anomalies.

Within the total patient group, we made an otologic distinction between major ear anomalies (at least meatal atresia) and minor ear anomalies (only ossicular chain and/or window malformations). Whenever possible, the ear anomaly was further classified according to Altmann,[10] Cremers et al,[11] and Cremers and Teunissen,[12] as shown in Table 2.[10-13] If relevant, pinna anomalies were classified according to Meurman.[13] The anatomic findings of the ears and the surgical technique applied (described in the operation reports or in the published reports) are presented schematically in the Tables.

The results of preoperative and postoperative audiological tests are presented as the pure tone average (PTA; mean of thresholds in decibels hearing level at 500, 1,000, and 2,000 Hz).

In the group with major ear anomalies, 4 of our patients did not undergo reconstructive surgery of the middle ear and aural canal, but they did undergo surgery for the purpose of fitting a bone-anchored hearing aid (BAHA). The anatomic details of the ears of these patients were obtained by means of radiodiagnostics using high-resolution computed tomographic scanning. Radiodiagnostic data were also available for the remaining patients, but as the methodology varied considerably over the 30-year period, these have been omitted here.

RESULTS

Of the 12 patients in our own material, 8 had major ear anomalies (Table 4, cases 1 through 4, and Table

TABLE 2. CLASSIFICATION OF MAJOR EAR ANOMALIES, MINOR EAR ANOMALIES, AND PINNA ANOMALIES

	Type	*Description*
Major ear anomalies[10,11]	I	Meatus is small and frequently only present in its medial portion
	IIA	Total bony atresia over only part of length of meatus, or canal is partially aplastic
	IIB	Total bony atresia over full length of meatus. Tympanic cavity may be smaller than normal
	III	Absent external meatus and small or missing tympanic cavity
Minor ear anomalies[12]	I	Isolated stapes ankylosis
	II	Stapes ankylosis with associated anomaly of incus and/or malleus
	III	Mobile stapes footplate, but anomalous or fixed incus and/or malleus
	IV	Aplasia or dysplasia of oval and/or round window
Pinna anomalies[13]	I	Pinna is smaller, rudimentary, and often located in abnormal position. Different parts of pinna are still discernible
	II	Pinna, besides being smaller and often in abnormal position, is represented by vertical curving ridge, resembling primitive helix
	III	Rudiment of pinna has no resemblance to any portion of normal pinna

TABLE 3. PATIENT DATA

Case	*Age* (y)*	*Sex*	*Authors*
1	16	M	This study
2	12	M	This study
3	10	F	This study
4	21	M	This study
5	16	M	This study
6	16	F	This study
7	18	M	This study
8	37	M	This study
9	5, 23	F	This study
10	33, 36	M	This study
11	7	F	This study
12	38	F	This study
13	1	F	Gill[23]
14	13	M	Gill[23]
15	7	F	Herberts[16]
16	19	M	Ombrédanne[20]
17	8	F	Fernandez and Ronis[18]
18	11	F	Fernandez and Ronis[18]
19	3	F	Fernandez and Ronis[18]
20	35	M	Cummings[19]
21	16	M	Plester[15]
22	12	F	Ombrédanne[22]
23	35	M	Edwards[17]
24	33	F	Fernandez and Ronis[18]
25	6	F	Fernandez and Ronis[18]
26	23	F	Gerhardt and Otto[24]
27	36	M	Holborow[14]
28	31	F	Holborow[14]
29	8	M	Fernandez and Ronis[18]
30	11	M	Fernandez and Ronis[18]
31	31	M	Herberts[16]
32	15	M	Ombrédanne[22]
33	8	M	Ombrédanne[21]

*At time of surgery.

9, cases 5 through 8). Case 3 involved asymmetric ear anomalies; the patient's left ear was classified in the major anomaly group and her right ear in the minor anomaly group. Four patients had bilateral minor ear anomalies (Table 6, cases 9 through 12). In the literature, only 11 of the 27 reports on ear surgery in relation to TCS described the individual patients in sufficient detail to be included in this study.[14-24] Tables 4, 5, 7, and 8 list the otologic findings, surgical techniques, and postoperative results of the patients described in the literature (cases 13 through 33, 21 patients and 28 ears). Patient-related items such as age at time of surgery and sex are summarized in Table 3.[14-24]

Major Ear Anomalies. The middle ear could not be identified during surgical exploration in 6 of the 14 ears with a major anomaly summarized in Table 4. Although a middle ear cavity was present in the remaining 8 ears, it was found that both the malleus and incus were either absent or seriously malformed, or fused and fixed in the epitympanum. The stapes was abnormal in 4 of the 7 reported ears, and in 2 ears it was absent. In cases 1, 4, and 19 a cholesteatoma was present.

Minor Ear Anomalies. The data on 25 ears with minor ear anomalies (Tables 5-8) showed that 14 ears had a type III minor ear anomaly: mobile (malformed) stapes but an anomalous or ankylotic incus and/or malleus (Fig 2). The malleus had a normal shape in only 4 of the 25 ears. In 3 cases, data on the malleus were lacking. The malleus was absent in 7 ears. Fusion between a malformed incus and malleus was found frequently, whether or not in combination with fixation in the epitympanum.

Only 1 patient had a normal incus (2 ears). In the majority of cases the incus was seriously malformed or even absent. Sometimes the incus appeared to have been replaced by a bony strand.

The stapes was abnormal in 24 of the 25 ears with minor ear anomalies (Fig 3). The stapes was monopodal in 7 ears (Fig 4). In 40% of the ears with minor anomalies, the stapes was fixed to the facial canal or was curved against the facial nerve. An ankylotic footplate was found in 28% of the minor ear anomalies.

Oval Window, Round Window, and Facial Nerve. In the two groups of patients with major or minor ear anomalies, the oval window was generally smaller than normal and there were 2 cases of aplasia. The round window was aplastic in 6 ears.

No records were available concerning the facial nerve in 8 of the 25 ears with minor anomalies. Only 3 of the 17 remaining ears had a normal facial nerve, and in many cases it was bare throughout the cavum tympani. In at least 7 ears, the oval window was partly covered by the facial nerve. Three of the cases with minor anomalies had an unusually thick chorda tympani.

Postoperative Hearing and Surgical Complications. Hearing improvement was achieved in only 5 of the 14 above-described ears with major anomalies. The average improvement was 30 dB (range, 20 to 40 dB); a postoperative PTA of less than or equal to 30 dB was found in 3 cases.

In 20 of the 25 ears with minor anomalies, there was a hearing improvement of at least 10 dB. The average hearing improvement in these 20 ears was 25 dB (range, 10 to 60 dB); a postoperative PTA of 30 dB

TABLE 4. OPERATIVE FINDINGS AND RESULTS IN PATIENTS WITH MAJOR EAR ANOMALIES

Case	*Ear*	*Type of Anomaly*	*Ossicular Chain*	*Oval and Round Windows*	*Facial Nerve*	*Type of Surgery*	*Remarks*	*Preoperative Hearing**		*Postoperative Hearing**	
								AC	*BC*	*AC*	*BC*
Nijmegen series											
1	L	I	Malleus and incus malformed, fixed in epitympanum, stapes mobile, bony stapedial tendon to incus	Normal	Normal	Canalplasty, myringoplasty, ossicular chain reconstruction with autologous ossicles	Congenital cholesteatoma	60	0	20	0
2	R	III	Middle ear cavity not identified					65	15	65	15
3	L	III	Middle ear cavity not identified					50	5	50	5
4	R	IIA	Malleus, incus, stapes fused into one rudimentary piece	Not reported	Bare, normal course	Mastoidectomy, tympanoplasty type IV	Congenital cholesteatoma	60	0	60	0
Literature											
13	R	III	Middle ear cavity not identified						Not reported		
	L	III	No malleus or incus found, stapes grossly deformed	Not reported	Deformed	Exploration			Not reported		
14	R	III	Middle ear cavity not identified						Not reported		
	L	III	Middle ear cavity not identified						Not reported		
15	?	II/III	Malformed caput malleus, manubrium absent, malformed incus, mobile stapes	Normal	Bare	Minor open cavity, tympanoplasty		65		40	
16	R	III	Absent, except rudimentary caput malleus	Aplasia	Not reported	Fenestration	Small middle ear cavity	70	15	50	15
	L	III	Atrophic, malformed incus, rudimentary caput malleus, stapes absent	Aplasia	Not reported	Fenestration		60	10	30	10
17	L	III?	Incus and malleus fused into one piece, stapes not reported	Not reported	Not reported	Mastoidectomy, tympanoplasty type III	No follow-up	55	5		
18	L	III	Middle ear cavity not identified						Not reported		
19	L	I	Malleus and incus absent, stapes deformed, no crura	Oval window covered by nerve VII	Bare	Mastoidectomy, fenestration, tympanoplasty type IV	Small attic cholesteatoma	65	5	30	

AC — air conduction, BC — bone conduction.

*Mean (decibels) of thresholds at 500, 1,000, and 2,000 Hz.

or less was found in only 10 ears. No hearing improvement was achieved in 5 of the 25 (20%) ears with type I, II, III, or IV minor anomalies.

When the patients were divided into groups on the basis of their age at surgery, it was found that the results of the patients who were older than 10 years (n

TABLE 5. OPERATIVE FINDINGS AND RESULTS OF PATIENTS WITH MINOR EAR ANOMALIES TYPES I AND II (SERIES FROM LITERATURE)

Case	*Ear*	*Type of Anomaly*	*Ossicular Chain*	*Facial Nerve*	*Type of Surgery*	*Remarks*	*Preoperative Hearing**		*Postoperative Hearing**	
							AC	*BC*	*AC*	*BC*
20	L	II	Fused malformed incus and malleus, caput of stapes fixed on processus cochleariformis, crura very delicate, ankylotic footplate	Normal	Stapedectomy, wire prosthesis from handle of malleus to vestibule	Chorda encased in bony plate	90	35	90	35
			Reexploration: extrusion of wire through eardrum, wire repositioned				90	35	45	35
21	R	I	Malleus and incus normal, monopodal ankylotic stapes fixed onto facial canal	Not reported	Stapes mobilization		55	0	15	0
	L	I	Malleus and incus normal, monopodal ankylotic stapes fixed onto facial canal	Not reported	Stapes mobilization		40	0	20	0
22	L	II	Short handle of malleus, fixed incus with delicate long process, monopodal ankylotic stapes	Bare	Stapedectomy, vein graft on oval window, autologous incus interpositioning to handle of malleus		60	0	30	10
23	R	II	Malleus not reported, long process of incus delicate and fibrous distally, monopodal stapes with restricted mobility	Not reported	Stapedectomy, vein graft on oval window, polyethylene strut (6 mm) to neck of malleus		65	10	25	10
24	R	II	Malleus and incus absent, stapes deformed, ankylotic footplate	Normal	Tympanoplasty type III		70	20	70	20
25	R	II	Malleus and incus absent, no stapes crura, ankylotic footplate	Bare	Exploratory tympanotomy only	Very large chorda tympani	60	0	60	0

AC — air conduction, BC — bone conduction.

*Mean (decibels) of thresholds at 500, 1,000, and 2,000 Hz.

= 13; mean hearing gain 23 dB; SD 18 dB) were better than those of the patients age 10 or younger (n = 6; mean hearing gain 9 dB; SD 8 dB); however, the difference was not significant (Student's $t = 1.78$; 17 *df*; $p > .05$).

The type of minor ear anomaly was not clearly related to the surgical result: types I and II showed a mean hearing gain of 17 dB (n = 6), whereas types III and IV were associated with a mean improvement of 18 dB (n = 14).

It was striking that none of the studies reviewed above mentioned a postoperative increase in the perceptive threshold or postoperative facial palsy, and similarly, none of the patients in our series suffered from these complications. Partly to avoid these complications, the operation was deemed unsuccessful and terminated in 20% of a total of 39 operated ears with anomalies.

Bone-Anchored Hearing Aid. Owing to the severity of the anomalies, we recently decided not to perform reconstructive surgery on 4 of our own patients with a major ear anomaly type III (cases 5, 6, 7, and 8). However, they were considered to be

TABLE 6. OPERATIVE FINDINGS AND RESULTS OF PATIENTS WITH MINOR EAR ANOMALIES TYPE III (NIJMEGEN SERIES)

Case	*Ear*	*Ossicular Chain*	*Facial Nerve*	*Type of Surgery*	*Remarks*	*Preoperative Hearing**		*Postoperative Hearing**	
						AC	*BC*	*AC*	*BC*
3	R	Malleus and incus fused, fixed in epitympanum, no caput mallei, long process malformed, rudimentary mobile stapes	Not reported	Ossicular chain reconstruction		50	10	35	5
9	R	Malformed malleus, incus absent, bony strand instead, rudimentary mobile stapes in contact with facial canal	Bare	Ossicular chain reconstruction with autologous cartilage	Marked thickening of chorda	50	5	40	5
		Reexploration: interpositioning of homologous transformed incus between stapes and chorda tympani				40	5	40	5
	L	Fixed, malformed malleus, rudimentary incus, stapes fixed onto facial canal, anterior crus absent	Bare	Mobilization of stapes, incus transpositioning between footplate and eardrum, malleus handle separated from fixed caput		55	0	40	0
		Reexploration: interpositioning of homologous incus between stapes and eardrum				50	5	30	5
10	R	Malleus normal, rudimentary incus with fragile long process fixed in epitympanum and onto facial canal, malformed mobile stapes	Normal	Autologous incus repositioning between stapes and eardrum		50	10	40	15
	L	Malleus normal but fixed, incus malformed and fixed, long process rudimentary and in contact with facial canal, stapes normal	Abnormal branch	Homologous corpus mallei interpositioning between stapes and eardrum, tympanoplasty	Thickening of chorda	40	20	25	5
11	L	Malleus and incus absent, bony strand between annulus and rudimentary stapes suprastructure, crura absent, mobile footplate	Bare, covering oval window	Exploratory tympanotomy, terminated		55	10	50	5
12	R	Malleus normal, incus absent, stapes curved against facial canal	Covering oval window	Homologous corpus mallei interpositioning between stapes and eardrum	Chronic otitis media	45	10	45	10

AC — air conduction, BC — bone conduction.

*Mean (decibels) of thresholds at 500, 1,000, and 2,000 Hz.

TABLE 7. OPERATIVE FINDINGS AND RESULTS OF PATIENTS WITH MINOR EAR ANOMALIES TYPE III (SERIES FROM LITERATURE)

					*Preoperative Hearing**		*Postoperative Hearing**	
Case	*Ear*	*Ossicular Chain*	*Facial Nerve*	*Type of Surgery*	*AC*	*BC*	*AC*	*BC*
26	R	Fused, rudimentary malleus and incus, stapes fragile and monopodal	Bare, partially covering oval window	Autologous incus-malleus interpositioning between footplate and eardrum	55	0	25	0
27	L	Malleus not reported, incus absent, thin strand between flattened stapes and malleus	Not reported	Polyethylene prosthesis interpositioning between stapes and eardrum	75	10	15	10
28	L	Malleus not reported, incus absent, stapes monopodal	Not reported	Polyethylene prosthesis interpositioning between stapes and eardrum	55	10	25	10
29	R	Malleus and incus malformed, malformed stapes fixed onto facial canal	Partially covering oval window	Polyethylene prosthesis interpositioning between stapes and tympanoplasty type III	65	0	Gain: nil	
	L	Malleus and incus absent, stapes deformed	Partially covering oval window	Polyethylene prosthesis interpositioning between stapes and tympanoplasty type III	45	5	Gain: nil	
30	R	Rudimentary malleus, incus malformed, malformed stapes against facial canal	Partially covering oval window	Tympanoplasty type III	65	10	45	
31	?	Absent caput mallei, malformed incus, absent footplate and crura	Not reported	Fenestration of posterior canal	60		45	

AC — air conduction, BC — bone conduction.
*Mean (decibels) of thresholds at 500, 1,000, and 2,000 Hz.

suitable candidates for a BAHA (Table 9) and underwent surgical implantation of a percutaneous titanium screw that was anchored in the temporal bone (Fig 5); at a later date the patients were fitted with the BAHA, which was attached to the implanted screw. The surgical procedures used have been described in detail by Tjellström.[25] The total follow-up period of these 4 patients was 80 months. Adverse skin reactions around the implant occurred 3 times, but could be treated successfully with antibiotic ointment. Two months after implantation, the patients underwent audiologic examinations to compare their performance with their previous conventional bone conduction hearing aid (CBHA) to their performance with the BAHA. The sound field warble tone threshold test showed lower thresholds with the BAHA, especially at the higher frequencies. The average threshold shift was –7 dB (range, +2 to –15 dB) per frequency (0.25 to 8 kHz). The sound field discrimination task in quiet could not differentiate between the two hearing aids, as the maximum phoneme score was 100% with both hearing aids in all patients. The speech-noise ratio with the Plomp test improved with the BAHA by an average of 2.1 dB (range, 0.5 to 4.4 dB), which is the equivalent of a 35% improvement in the speech score in noise.[26] In 2 of the 4 patients, this improvement was statistically significant.

DISCUSSION

Major Ear Anomalies. Atresia of the aural canal in patients with TCS, in combination with dysplasia of the pinna, forms a serious functional and cosmetic problem. Meatal atresia is found in 36% of the affected persons.[27] This percentage is probably on the high side, because the variation in expression of TCS has so far been underestimated.[28] The majority of patients have a type III anomaly, and radiographic examinations nearly always show the absence of a middle ear, which represents the most complicated major ear anomaly. Besides meatal atresia, TCS also has other characteristic abnormalities, such as the absence of mastoid pneumatization, an anteriorly placed mastoid segment and consequently an anteriorly placed sigmoid sinus, a low-lying tegmen, and, in accordance with the classification of type III, a slitlike or absent middle ear cavity.[29,30] However, the mesotympanum and hypotympanum are more nor-

TABLE 8. OPERATIVE FINDINGS AND RESULTS OF PATIENTS WITH MINOR ANOMALIES TYPE IV (SERIES FROM LITERATURE)

Case	*Ear*	*Ossicular Chain*	*Facial Nerve, Oval and Round Windows*	*Type of Surgery*	*Preoperative Hearing**		*Postoperative Hearing**	
					AC	*BC*	*AC*	*BC*
22	R	Fused malleus and incus, rudimentary caput mallei, incus fixed in aditus, long process absent, malformed stapes crura, ankylotic footplate	Bare facial nerve, partially covering normal oval window, aplasia of round window	Stapedectomy with vein graft on oval window and reinterpositioning of stapes, creation of neo–round window	55	10	20	10
32	L	Malleus absent, corpus incudis rudimentary, stapes bent over facial nerve, small footplate	Facial nerve not reported, small oval window, aplasia of round window	Stapedectomy, creation of neo–round window, interpositioning of bone chip	65	25	50	25
33	R	Malleus absent, incus malformed, long process absent, minuscule stapes remnant partially covered by facial nerve	Bare facial nerve covering small oval window, round window absent	Stapedectomy, creation of neo–round window, tympanoplasty type IV with skin graft	75	15	55	15
	L	Absent malleus and incus, minuscule stapes remnant partially covered by facial nerve	Bare facial nerve, small oval window, round window absent	Stapedectomy, creation of neo–round window, tympanoplasty type IV	60	25	40	25

AC — air conduction, BC — bone conduction.

*Mean (decibels) of thresholds at 500, 1,000, and 2,000 Hz.

mal than the epitympanum (especially if the external meatus is present).[31] Recent studies have shown a bony cleft in the lateral aspect of the temporal bone just anterior to the mastoid.[9] This finding may be clinically relevant, because it is likely that the cleft contains the facial nerve, which generally runs an abnormal and more anterior course from the geniculate ganglion. Sometimes dehiscence of the canal in the

TABLE 9. COMPUTED TOMOGRAPHIC SCAN FINDINGS OF FOUR PATIENTS WITH MAJOR ANOMALIES WHO WERE FITTED WITH BONE-ANCHORED HEARING AID (NIJMEGEN SERIES)

Case	*Ear*	*Type of Pinna Anomaly*	*Type of Meatus Anomaly*	*Mastoid*	*Middle Ear Cavity and Ossicles*	*Facial Nerve*	*Inner Ear, Round and Oval Windows*	*Hearing Threshold**	
								AC	*BC*
5	R	I	IIA	No pneumatization, hypoplastic	Aerated, slitlike cavity; rudimentary ossicles in epitympanum	Aberrant course of descending part	Normal	65	5
	L	I	IIA	Symmetry				55	0
6	R	III	III	No pneumatization, severe hypoplasia	Very narrow, slitlike cavity, filled with soft tissue; fixed, rudimentary ossicles	Aberrant course of descending part	Small lateral semicircular canal, wider than normal	75	15
	L	III	III	Not reported				75	5
7	R	II	III	No pneumatization, hypoplastic	Aerated, slitlike cavity; ossicles absent	Aberrant course of descending part	Normal	60	0
	L	III	III	Symmetry				55	5
8	R	II	IIA	No pneumatization, hypoplastic	Aerated, slitlike cavity; ossicles absent	Not visualized	Not visualized	75	15
	L	II	I	Symmetry				70	10

AC — air conduction, BC — bone conduction.

*Mean (decibels) of thresholds at 500, 1,000, and 2,000 Hz.

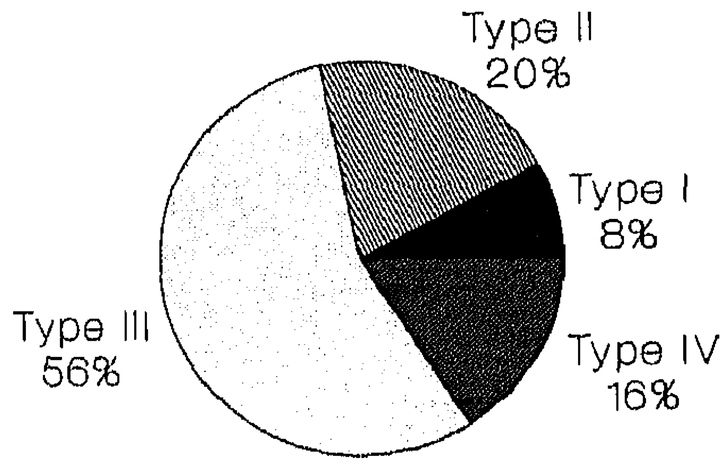

Fig 2. Overview of type of middle ear anomaly as defined in Table 2 of 25 ears with only congenital middle ear anomaly in patients with Treacher Collins syndrome (total series).

middle ear is observed.

Although not typical for TCS, a congenital cholesteatoma was found in 3 ears of a total of 14 operated ears with major anomalies. Two of these ears had bony atresia. This finding is not unusual in cases with congenital aural atresia. In a series of 62 ears with partial or total atresia, 8% were found to have a cholesteatoma.[32] However, in our own patients, this never formed the primary indication for surgery. There is no evidence in the literature that the presence of a congenital cholesteatoma without symptoms should form a clear indication for surgery within this patient group.

Computed tomography makes it possible to classify aural atresia and now forms an essential part of the preoperative diagnostic procedure.

The abnormalities in TCS patients with major ear anomalies are so complex that in our opinion there is seldom a good indication for reconstructive surgery. This is particularly the case with type II and type III (aural atresia) anomalies. However, our own experience and that of other authors has shown that it is possible to achieve positive results in selected cases. In our opinion, there is a better alternative for the majority of patients nowadays, as they can be fitted with a CBHA or a BAHA. With the CBHA, problems are frequently encountered in relation to the fitting of the aid, the cosmetic appearance, skin irritation as a result of the constant pressure applied to the mastoid

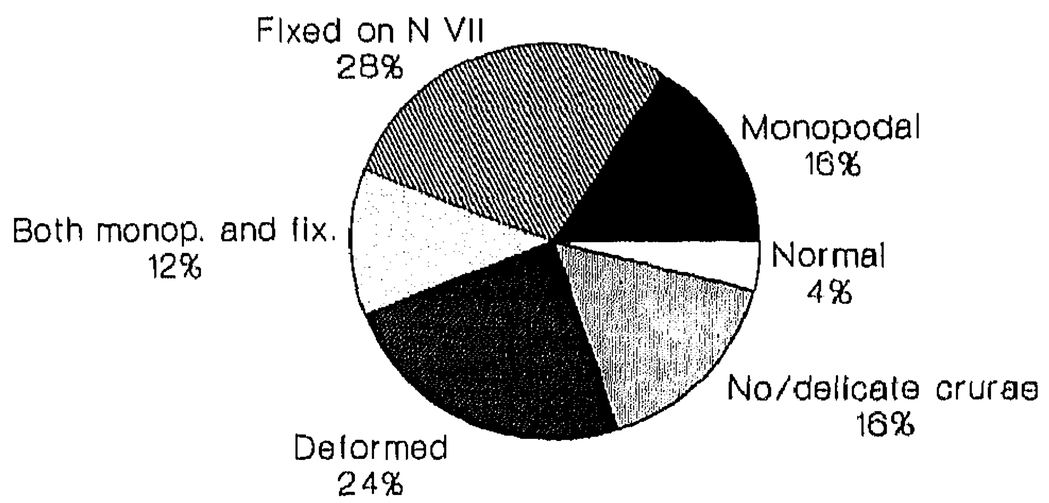

Fig 3. Description of stapes anomalies in 25 ears with only congenital middle ear anomaly (total series).

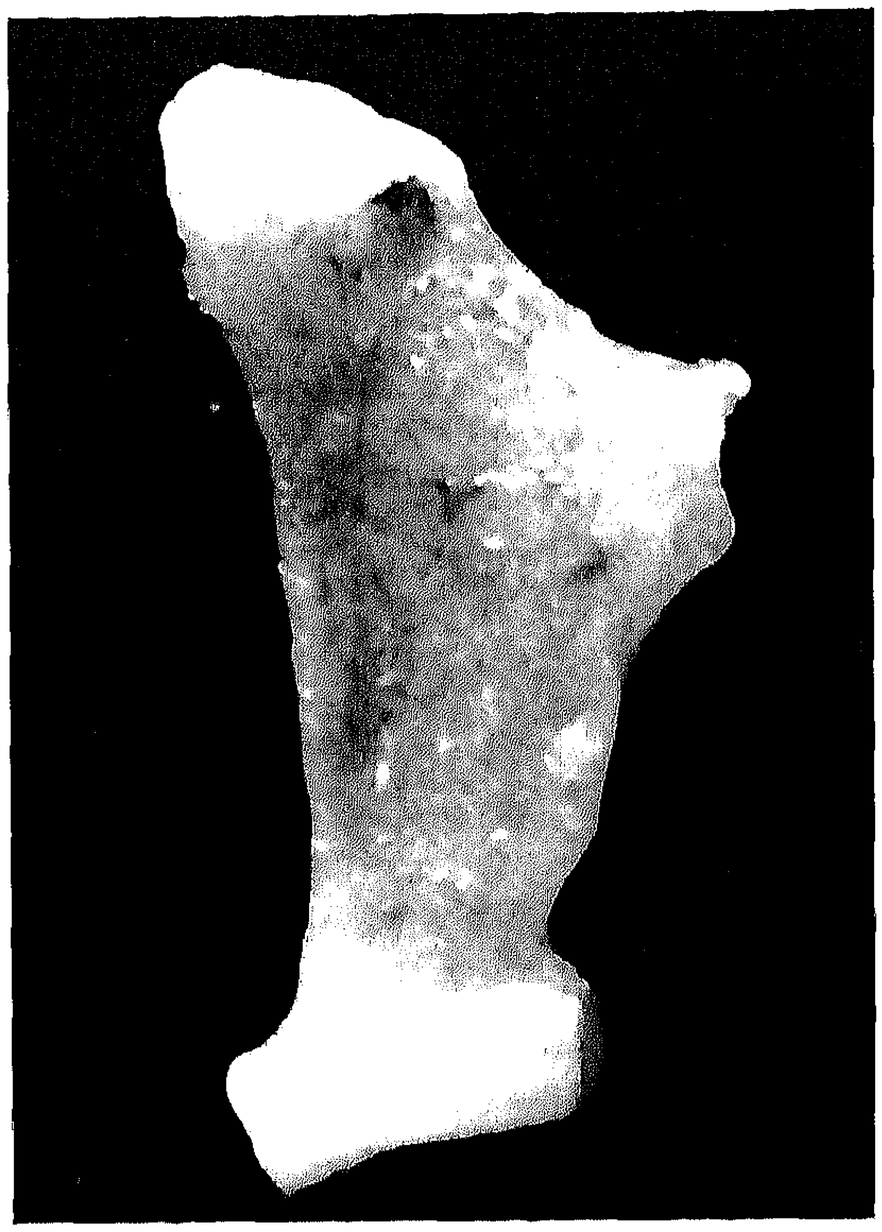

Fig 4. Monopodal stapes in Treacher Collins syndrome, including whole footplate (bottom).

process, and the lack of support for the hearing aid owing to microtia, as was the case in patients 6 and 7 (Table 9). Four of our patients were fitted with a BAHA because they experienced these problems with the CBHA. All the patients can wear their new

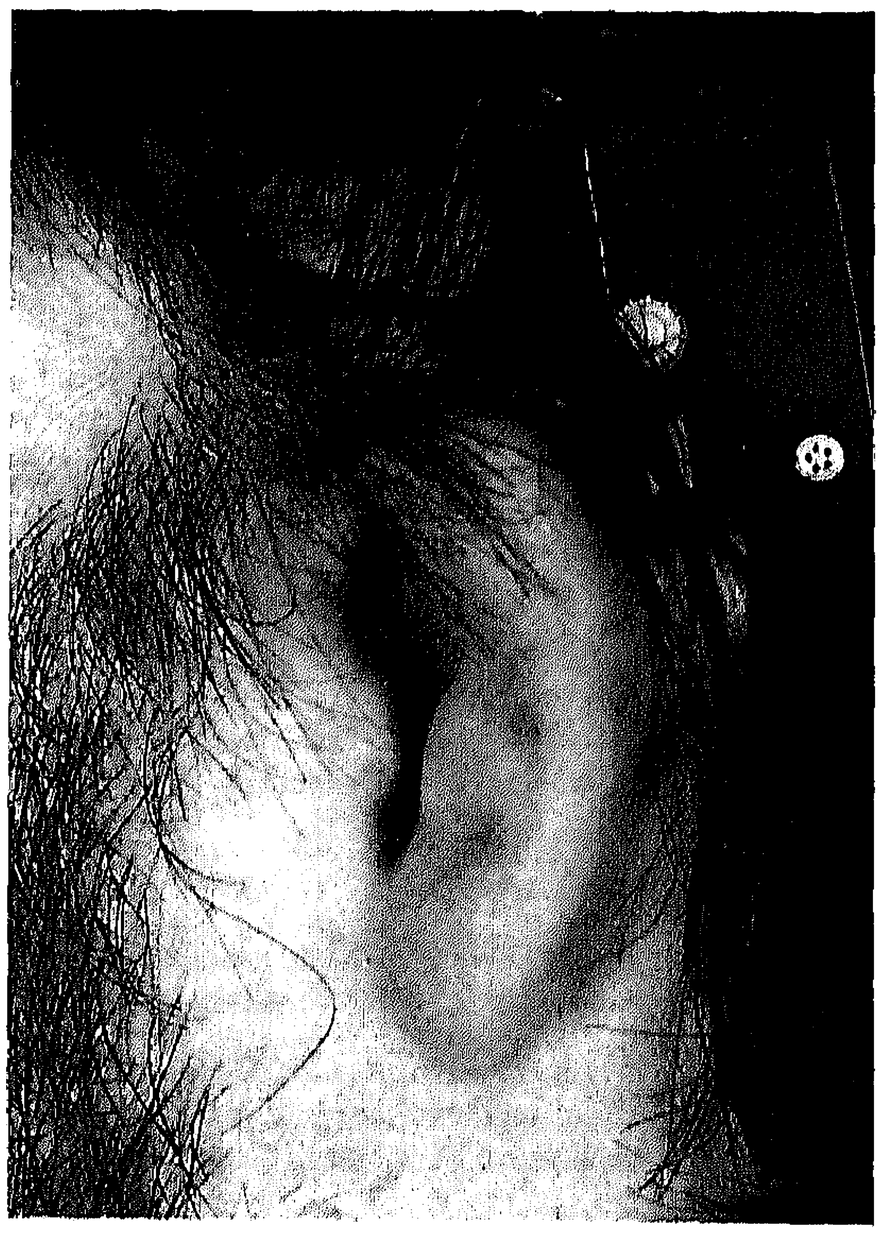

Fig 5. (Case 8) Bone-anchored hearing aid.

hearing aid all day and prefer it to the CBHA. The most important advantages reported by the patients are better speech intelligibility in noise and an improved aesthetic appearance.

Minor Ear Anomalies. If a patient presents with a congenital anomaly of the middle ear only, reconstructive surgery can be considered if he or she is older than 10 years of age.[12] The preoperative diagnostics comprise otoscopy, pure tone and speech audiometry, tympanometry, and high-resolution computed tomographic scanning of the temporal bones to evaluate the middle ear, its contents, and the surrounding structures. Besides the necessary experience of the otologist, the anesthetist should also have experience with the specific perioperative complications within this patient group, such as potential problems with airway management.[33]

The surgical findings that can be expected during exploratory tympanotomy are diverse, such as the absence of (part of) the ossicular chain, severe deformity of the ossicular chain, ankylosis of the malleus and/or incus and/or stapes footplate, a monopodal stapes, an oval window partially covered by the facial nerve, and close contact between the stapes and the facial nerve.

The disappointing results of reconstructive surgery in TCS patients are generally caused by a deformed stapes. If the stapes is mobile but the stapes head is bent toward the facial nerve, this will impede successful ossicular chain reconstruction. We no longer perform stapes mobilization in the case of ankylosis of the footplate or fixation to the facial canal, in view of the poor long-term follow-up results. In these situations, new techniques should be applied. One such technique involves making a stapedotomy opening in the footplate and subsequently removing the stapes suprastructure by means of an argon laser. Functional ossicular chain reconstruction can be achieved by malleovestibulopexy, as described by Edwards[17] and Cummings.[19] Fixation of a piston to the chorda can be carried out if the malleus handle is absent, and then myringochordovestibulopexy can be created by interpositioning cartilage between the chorda and the tympanic membrane.[34] Whether these new techniques lead to the desired level of hearing gain should become clear in the near future.

CONCLUSIONS

The early detection of (possible) hearing loss in TCS patients is of great importance in order to be able to rehabilitate their hearing with a hearing aid. Classifying an ear with aural atresia has proved to be worthwhile so that it can be estimated whether reconstructive ear surgery will be sufficiently successful. Computed tomography of the ear is a necessity in this respect. The diagnosis of TCS in combination with congenital aural atresia appears to predict fairly frequently that the degree of malformation of the middle ear will be so severe that reconstructive surgery of the middle ear function will only be successful in exceptional cases. Rehabilitation with a CBHA or a BAHA generally takes precedence over reconstructive ear surgery.

Similarly, if the patient has a minor ear anomaly, the hearing loss will be so severe that rehabilitation should be started at the earliest possible opportunity with an air conduction hearing aid. The patient's hearing can be improved surgically, but it is best to delay the operation until he or she has reached the age of 10 years. The chances of success are lower than usual because of the presence of associated anomalies of the ossicular chain. In addition, patients with TCS have specific abnormalities of the stapes that impede successful reconstruction of the ossicular chain. The facial nerve often runs a deviant course that involves the risk of damage during surgery. The application of the new surgical techniques requires extra expertise from the surgeon.

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