Клинические сведения
Синдром Тричера-Коллинза и Baha®

Система Cochlear™ Baha® 3

Синдром Тричера-Коллинза (СТК) (синдром Франческетти или челюстно-лицевой дизостоз) – генетическое заболевание, характеризующееся деформацией лицевого черепа. Для этого синдрома типично недоразвитие скуловых костей, верхней и нижней челюстей, что приводит к деформации орбит и век. У пациентов с синдромом Тричера-Коллинза часто наблюдается хроническая дыхательная недостаточность, апноэ во время сна и кондуктивная тугоухость, обусловленная пороками развития наружного и среднего уха[1]. Применение системы Baha позволяет преодолеть кондуктивную тугоухость, обусловленную врожденными пороками развития уха.

ЧАСТОТА

Дети с синдромом Тричера-Коллинза рождаются редко – 1 случай на 10 000 новорожденных[1,2]. Приблизительно у 30% детей с этим синдромом есть расщелина неба, и у большинства таких детей выявляется кондуктивная тугоухость умеренной или тяжелой степени[3].

ТУГОУХОСТЬ

Тугоухость при синдроме Тричера-Коллинза обусловлена нарушениями строения наружного и среднего уха. Аномалии развития уха делятся на малые (недоразвитие структур наружного и среднего уха) и большие (частичное или полное отсутствие наружного и среднего уха). Пороки развития среднего уха выявляются у большинства детей с синдромом Тричера-Коллинза, в то время как пороки развития внутреннего уха с сенсоневральной тугоухостью для этого синдрома нехарактерны.

Тугоухость, как правило, двусторонняя, с кондуктивным снижением слуха примерно на 50–70 дБ. Даже у детей с нормально сформированными ушными раковинами и открытыми наружными слуховыми проходами часто отмечается деформация слуховых косточек[1].

ПОЛЬЗА ОТ УСИЛЕНИЯ ЗВУКА

Пациентам с синдромом Тричера-Коллинза требуется многопрофильный подход к оказанию медицинской помощи с привлечением медицинских работников разных специальностей[4]. такие дети нуждаются не только в проведении множественных хирургических вмешательств, направленных на реконструкцию лицевого черепа. Снижение слуха и нарушения речи крайне негативно сказываются на способностях к обучению, самооценке и на формировании навыков социального общения[5]. Сообщалось о случаях задержки речевого развития, связанной с отсутствием речевой стимуляции на фоне тугоухости в первые годы жизни. Даже наличие кондуктивной тугоухости легкой степени замедляет формирование навыков устной и письменной речи и уменьшает разборчивость слов, произносимых ребенком[1]. У таких пациентов очень важно рано выявлять тугоухость и корригировать слух; крайне желательно, чтобы это было сделано до достижения ребенком 6-месячного возраста[5].

ЛЕЧЕНИЕ ТУГОУХОСТИ

Попытки хирургической реконструкции наружного слухового прохода для улучшения слуха у детей с синдромом Тричера-Коллинза оказались неэффективными[6]. Результатами клинических исследований было доказано, что восстановление слуха с помощью системы Baha или обычного слухового аппарата костной проводимости предпочтительнее хирургической реконструкции[7].
У пациентов с синдромом Тричера-Коллинза применение аппарата Baha характеризуется несколькими преимуществами:

- Поскольку раннее слухопротезирование крайне важно для таких пациентов, использование звукового процессора Baha на эластичной ленте Baha Softband способствует нормальному развитию речи[8]
- При использовании звукового процессора Baha на эластичной ленте Baha Softband у детей с пороками развития лицевого черепа регистрировалось улучшение порогов слышимости при тональной аудиометрии в речевом диапазоне >40 дБ[9]
- По сравнению с обычными слуховыми аппаратами костного звукопроведения при применении системы Baha результаты лучше – как с аудиологической, так и с эстетической точки зрения[10]
- По сравнению с хирургической реконструкцией при применении системы Baha аудиологические результаты намного лучше, а конечные исходы лечения более предсказуемы[6]
- После установки системы Baha возможны спонтанное улучшение качества речи, высоты и интенсивности собственного голоса пациента[11]

ЗАКЛЮЧЕНИЕ

Применение системы Baha позволяет преодолеть кондуктивную тугоухость, обусловленную пороками развития уха при синдроме Тричера-Коллинза. Сообщалось о существенном прогрессе развития речи у пациентов с синдромом Тричера-Коллинза после установки системы Baha. Кроме того, использование эластичной ленты Baha Softband дает возможность раннего слухопротезирования, которое крайне важно для этой группы пациентов[8,9].

СПИСОК ЛИТЕРАТУРЫ

1. Argenta LC, Iacobucci JJ. Treacher Collins syndrome: present concepts of the disorder and their surgical correction. World journal of surgery. 1989 Jul-Aug;13(4):401-9.

Treacher Collins Syndrome is a rare bilateral congenital deformity occurring in 1 in 10,000 births. It is also known, in the European literature, as Franceschetti Syndrome, and is additionally known as mandibulofacial dysostosis. It is a syndrome with a very wide spectrum of manifestations characterized by distortions of the orbit secondary to hypoplasia of the maxilla, mandible, and, most markedly, of the zygoma. Soft tissue deformities include lower lid colobomas, laxity and dystopia of the lateral canthus, microtia, and a paucity of the muscular aponeurosis of the midface. The syndrome is frequently accompanied by significant hearing loss, early failure to thrive, chronic respiratory insufficiency, and sleep apnea. Intelligence is usually within normal limits although learning disabilities are common in early life. These major anatomical and physiological abnormalities, as well as the psychological and social stigma associated with severe facial deformity, make this syndrome one of the most challenging reconstructive problems presented to the craniofacial surgeon.

2. Trainor PA, Dixon J, Dixon MJ. Treacher Collins syndrome: etiology, pathogenesis and prevention. European journal of human genetics. 2009 Mar;17(3):275-83.

Treacher Collins syndrome (TCS) is a rare congenital disorder of craniofacial development that arises as the result of mutations in the TCOF1 gene, which encodes a nucleolar phosphoprotein known as Treacle. Individuals diagnosed with TCS frequently undergo multiple reconstructive surgeries, which are rarely fully corrective. Identifying potential avenues for rescue and/or repair of TCS depends on a profound appreciation of the etiology and pathogenesis of the syndrome. Recent research using animal models has not only determined the cellular basis of TCS but also, more importantly, unveiled a successful avenue for therapeutic intervention and prevention of the craniofacial anomalies observed in TCS.

3. Pron G, Galloway C, Armstrong D, Posnick J. Ear malformations and hearing loss in patients with Treacher Collins Syndrome. Cleft Palate Craniofacial Journal. 1993 Jan 30,1:97-103.

Although the hearing loss of patients with Treacher Collins syndrome is well documented, few studies have reported jointly on their hearing loss and ear pathology. This paper reports on the hearing loss and computerized tomography (CT) assessments of ear malformations in a large pediatric series of patients with Treacher Collins. Of the 29 subjects assessed by the Craniofacial Program between 1986 and 1990, paired audiologic and complete CT assessments were available for 23 subjects. The external ear canal abnormalities were largely symmetric, either bilaterally stenotic or atretic. In most cases, the middle ear cavity was bilaterally hypoplastic and dysmorphic, and ossicles were symmetrically dysmorphic or missing. Inner ear structures were normal in all patients. The majority of patients had a unilateral or bilateral moderate or greater degree of hearing loss and almost half had an asymmetric hearing loss. The hearing loss of all subjects was conductive, except for three whose loss was bilateral mixed. Two types of bilaterally symmetric hearing loss configurations, flat and reverse sloping, were noted. Conductive hearing loss in patients with Treacher Collins is mainly attributable to their middle ear malformations, which are similar for those of patients with malformed or missing ossicles.

4. Reuter WF, Marks C, Jovanovic S, Gross M. Rehabilitation in Fraceschetti Syndrome: An interdiciplinary approach using bone-anchored hearing aids. ENT-Ear, Nose & Throat Journal. 1997 ; 76 (6) 402-10.

The purpose of the study was to determine the effectiveness of a concept of combined interdisciplinary rehabilitation for children with mandibulofacial dysostosis, developed at the Center for Facial Malformations. It consists of binaural implantation of bone-anchored hearing aids and gradual distraction of the mandible. After audiological testing and mandibular distraction on a phantom head designed with data from a spiral CT, the surgery was done in three steps: implanting the fixtures for Baha and the bone-lengthening device, removing the device after six weeks and completing the Baha implantation two months later. The distraction procedure and orthodontic treatment were performed on an outpatient basis. The results (six patients, ages 6-19 years) were excellent: after implantation of the Baha system speech perception increased from approximately 85% with the conventional BCHA to 95-100% with the Baha. Quality of life was reported to be much better because of the general cosmetic improvement as well as the good acoustic orientation and sound quality with the new hearing devices. We conclude that the interdisciplinary approach provides favorable conditions for rehabilitation in cases of complex malformations of the head and neck.

5. Yoshinaga-Itano C. Early intervention after universal neonatal hearing screening: impact on outcomes. Mental retardation and developmental disabilities research reviews. 2003;9 252-266.

This article summarizes the developmental outcomes of Colorado children with significant hearing loss. Some of the research compares children born in hospitals that have implemented universal newborn hearing screening programs for newborns. Other research compares the developmental outcomes of children who have been early-identified with hearing loss. Early-identification is defined as identification of hearing loss within the first six months of life. Late identification in the Colorado studies is defined as age of identification of hearing loss after the age of six months. In a few of the Colorado studies, age at initiation of intervention was used. Within the Colorado system, age of identification can be interpreted as almost synonymous with age of intervention, as the vast majority of children enter intervention services with two months after the identification of the hearing loss. Children who were early-identified and had early initiation of intervention services (within the first year of life) had significantly better vocabulary, general language abilities, speech intelligibility and phoneme repertoires, syntax as measured by mean length of utterance, social-emotional development, parental bonding, and parental grief resolution. Two other studies (Nebraska and Washington state) of early- versus later-initiation of intervention services report findings similar to the Colorado studies. Direct comparisons with the historical literature are not possible because the developmental delays of what would now be termed “later-identified” were too low to report developmental ages for the birth through five-year-old population.

6. Marres HA, Cremers CW, Marres EH. The Treacher Collins syndrome. Management of major and minor ear anomailes. Revue de laryngologie otololologie rhinologie. 1995:116, 2;105-108.

12 patients suffering from a Treacher Collins syndrome, or mandibulo-facial dysostosis, were operated on in the Nijmegen University Hospital between 1960 and 1990. An early diagnosis is generally reached when there is a congenital atresia of the auditory canal. Auditory rehabilitation with a conventional prosthesis of the bone or a Baha is preferable to surgical reconstruction. In minor cases, deafness must be screened as early as possible, with a bone hearing aid prosthesis. Surgical exploration can be performed at best as the age of 10, but the chances of success are less than those of other functional reconstructions because of the associated malformation.

7. Marres HA. Hearing Loss in the Treacher Collins Syndrome. Adv otorhinolaryngol. 2002;61:209-15.

About 50% of patients with the TCS have conductive hearing loss, caused by characteristic major and/or minor ear anomalies. It is also common for these patients to have microtia or severe malformation of the pinna. The results of [figure: see text] reconstructive surgery to improve hearing are usually moderate to poor, because of combinations of anomalies. In general, it is recommended to start hearing rehabilitation at the earliest possible stage. Owing to the above-described anomalies, rehabilitation usually involves fitting a Baha, whether or not in combination with a pinna epithesis.

8. Verhagen CV, Hol MK, Coppens-Schellekens W, Snik AF, Cremers CW. The Baha Softband A new treatment for young children with bilateral congenital aural atresia. International Journal of Pediatric Otorhinolaryngology (2008) 72, 1455—1459.

The Baha (Bone-Anchored hearing aid) Softband appears to be an effective mean of hearing rehabilitation for children with a congenital bilateral aural atresia who are too young for the amplification of a Baha on an implant. The aided hearing threshold with a Baha Softband is almost equal to that achieved with a conventional bone conductor. The speech development of the children studied with a Baha Softband is on a par with peers with good hearing.

9. Nicholson N, Christensen L, Dornhoffer J, Martin P Smith-Olinde L. Verification of speech spectrum audibility for pediatric Baha Softband users with craniofacial anomalies. Cleft palate craniofacial journal. 2011 Jan 48,1

Objective: The purpose of this study was (1) to determine benefit of the Baha Softband coupled to the Softband for infants and children with bilateral conductive hearing loss; and (2) to verify audibility of the speech spectrum for octave frequencies 500 through 4000 Hz. Design: The research design for this retrospective chart study is pretest-posttest repeated measures. Setting: The study was conducted in the Department of Audiology and Speech Pathology, Arkansas Children's Hospital. Participants: 25 children aged six months to 18 years with craniofacial disorders and bilateral conductive hearing loss participated in the study. Participants were consistent, full-time unilateral Baha users with the Baha Compact bone-conduction amplifier coupled to the head via the Softband. Interventions: The intervention was the Baha device coupled to the head via the Softband as a prerequisite to surgical implantation. Main Outcome Measure(s): The primary study outcome measures used aided and unaided soundfield audiometric thresholds to calculate functional gain. Audibility of the speech spectrum was verified by comparison with target aided thresholds. Results: Results revealed an improvement in soundfield thresholds with Baha amplification for the four octave frequencies. Means, standard deviations, and confidence intervals for aided and unaided thresholds are reported. Percentages of thresholds meeting target levels were significant at all frequencies, exceeding the 80% criterion. Conclusions: Benefit of the Baha in providing audibility of the speech spectrum for infants and children with bilateral congenital conductive hearing loss has been demonstrated, offering important and timely data supporting third-party reimbursement.

10. Granström G, Tjellström A. The Bone Anchored hearing aid in children with auricular malformations. ENT.Ear Nose Throat journal. 1997.

A retrospective study was undertaken to evaluate the outcome of the use of the bone-anchored hearing aid (Baha) in children. All patients included in the study had bilateral auricular malformations. Previous alternatives had been conventional hearing aids or surgical middle ear reconstruction. 37 patients under 16 years of age were studied. The most common syndrome in the group was Treacher Collins. 16 of the patients had earlier middle ear reconstruction, the results of which did not produce social hearing. Of 40 inserted fixtures to anchor the Baha, three were lost during the follow-up period because of failed osseointegration. Skin reactions were graded according to a clinical scoring system and were determined to be comparable in number and severity to those of an adult population. All patients in the study considered the Baha to be superior to earlier bone-conduction devices. It is concluded that the Baha is an excellent alternative to bone-conduction devices in children with auricular malformations. Middle ear surgery can be postponed until adulthood or abandoned, especially in syndromic patients in whom it is known to be difficult and unpredictable.

11. Thomas J. Speech and voice rehabilitation in selected patients fitted with a bone anchored hearing aid (Baha). The journal of laryngology and otology. Supplement. 1996;21:47-51.

With the Birmingham osseointegrated implant programme there have been several patients with severe pre-lingual conductive hearing loss. The majority of these have been patients with Treacher Collins syndrome. There are characteristic features of speech and voice in those with long-standing conductive hearing loss. In addition, the associated abnormalities of jaw, teeth and palate may amplify the problem. There may be spontaneous improvement in features such as voice pitch, quality and intensity following the fitting of a Baha. However, in those with a pre-lingual hearing impairment, speech therapy may be necessary. Patients assessed as suitable for Baha have a full assessment of communication skills including audio recording of speech and voice. Post-operative training improves auditory discrimination and perception and is followed by training in the production of the newly perceived speech sounds.

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