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Wady wrodzone ucha wewnętrznego - nowoczesna metoda obrazowania tomografią komputerową i jej znaczenie w audiologii

Rozprawa na stopień doktora nauk medycznych i nauk o zdrowiu w dyscyplinie nauki medyczne

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Tomografia komputerowa
Niedosłuch
Implant ślimakowy
Przewód słuchowy wewnętrzny
Kanał półkolisty
Zespół CHARGE
Słuchowe potencjały wywołane pnia mózgu
Słuchowe potencjały wywołane stanu ustalonego

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1. **Szleper A**, Lachowska M, Pastuszka A, Łukaszewicz-Moszyńska Z, Wojciechowski T, Niemczyk K. Anatomical and clinical aspects and outcomes of bilateral cochlear implantation in cochlear hypoplasia type IV – a case report. Polski Przegląd Otorynolaryngologiczny. (2022);11(4):57-63. DOI: 10.5604/01.3001.0016.2238

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2. **Szleper A**, Lachowska M, Wojciechowski T, Niemczyk K. Computed tomography multiplanar and 3D image assessment protocol for detailed analysis of inner ear malformations in patients undergoing cochlear implantation counseling. Otolaryngologia Polska. 2024;78(2):35-43. DOI: 10.5604/01.3001.0054.2567

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Wykaz stosowanych skrótów

CT	tomografia komputerowa (ang. *comupted tomography*)
ABR	słuchowe potencjały wywołane pnia mózgu (ang. *auditory brainstem response*)
NRT	telemetria odpowiedzi neuronowej (ang. *neural response telemetry*)
ASSR	słuchowe potencjały wywołane stanu ustalonego (ang. *auditory steady-state response*)
CADV	aplazja ślimaka z poszerzonym przedsionkiem (ang. *cochlear aplasia with a dilated vestibule*)
IAC	przewód słuchowy wewnętrzny (ang. *internal acoustic canal*)
HU	jednostki Hounsfielda (ang. *Hounsfield units*)

Streszczenie w języku polskim

Wstęp

Wady wrodzone ucha wewnętrznego powstające w trakcie embriogenezy mogą być uwidocznione w badaniach obrazowych. Patologie te stanowią ok 20% przyczyn wrodzonego niedosłuchu zmysłowo-nerwowego i charakteryzują się dużym zróżnicowaniem anatomicznym oraz szerokim spektrum zaawansowania klinicznego, w tym audiologicznego. Wraz z postępem technologicznym coraz bardziej precyzyjne obrazowanie za pomocą tomografii komputerowej (CT) o wysokiej rozdzielczości oraz badania rezonansu magnetycznego umożliwiły stworzenie kilku klasyfikacji stosowanych w praktyce klinicznej. Najbardziej kompleksową klasyfikacją wad rozwojowych ucha wewnętrznego jest klasyfikacja Sennaroğlu, opracowana po raz pierwszy w 2002 roku, a następnie zmodyfikowana w 2017 roku w celu uwzględnienia najnowszych odkryć.

Prawidłowa identyfikacja wady wrodzonej ucha ma kluczowe znaczenie dla dostosowania odpowiedniej metody rehabilitacji słuchu. Niektóre z tych wad można skorygować za pomocą aparatów słuchowych, podczas gdy inne, takie jak głęboki obustronny niedosłuch odbiorczy, wymagają wszczepienia implantu ślimakowego.

Cele

1. Opracowanie metodologii rekonstrukcji wielopłaszczyznowej obrazów tomografii komputerowej kości skroniowych pacjentów z wadami wrodzonymi ucha wewnętrznego uzupełnionej rekonstrukcją 3D .
2. Zastosowanie opracowanego protokołu rekonstrukcji i analizy obrazów tomografii komputerowej ucha wewnętrznego w praktyce klinicznej w procesie kwalifikacji do implantacji ślimakowej pacjentów z wadami ucha wewnętrznego.
3. Ocena, przy użyciu wypracowanego protokołu, wad wrodzonych ucha wewnętrznego u pacjentów z zespołem CHARGE oraz korelacja wyników analizy zrekonstruowanych obrazów tomografii komputerowej ucha wewnętrznego z wynikami badań audiologicznych.

Publikacja #1

Szleper A, Lachowska M, Pastuszka A, Łukaszewicz-Moszyńska Z, Wojciechowski T, Niemczyk K. Anatomical and clinical aspects and outcomes of bilateral cochlear implantation

in cochlear hypoplasia type IV – a case report. Polski Przegląd Otorynolaryngologiczny. (2022);11(4):57-63. DOI: 10.5604/01.3001.0016.2238.

Publikacja pierwsza rozpoczyna cykl analiz badań obrazowych, w tym przypadku rozszerzony o metodę rehabilitacji narządu słuchu oraz jej wstępne wyniki. W opisywanym przypadku, u 6-miesięcznego pacjenta, zdiagnozowano obustronną wadę rozwojową ucha wewnętrznego – hipoplazję ślimaka typu IV. W celu szczegółowej analizy anatomii ucha środkowego i wewnętrznego przeprowadzono rekonstrukcję wielopłaszczyznową obrazu tomografii komputerowej oraz rekonstrukcję 3D. Obie techniki rekonstrukcji obrazowej były bardzo pomocne w zindywidualizowanym podejściu do leczenia opisywanej pacjentki .

Publikacja #2

Szleper A, Lachowska M, Wojciechowski T, Niemczyk K. Computed tomography multiplanar and 3D image assessment protocol for detailed analysis of inner ear malformations in patients undergoing cochlear implantation counseling. Otolaryngologia Polska. 2024;78(2):35-43. DOI: 10.5604/01.3001.0054.2567.

Praca stanowi zaprezentowanie protokołu rekonstrukcji wielopłaszczyznowej obrazów CT ucha wewnętzrznego uzupełnionych o rekonstrukcje 3D, co pomaga klinicystom w lepszym zrozumieniu istoty danej wady oraz usprawnia podejmowanie decyzji terapeutycznych. Praca stanowi szczegółowe przedstawienie schematów analizy badań CT zaproponowanych w pierwszym artykule cyklu. Grupę badawczą stanowili pacjenci poddawani diagnostyce obrazowej oraz audiologicznej w toku kwalifikacji do wszczepienia implantu ślimakowego. Retrospektywnej analizie poddano 11 pacjentów (22 uszu) z wykrytą wadą wrodzoną ucha wewnętrznego. Wyniki przedstawiono w formie licznych, czytelnych wizualizacji uzupełniono o dane opisowe, co pozwoliło na dokładniejsze zaprezentowanie wyników badania.

Publikacja #3

Szleper A, Lachowska M, Wojciechowski T, Pronicka-Iwanicka K. Detailed analysis of inner ear malformations in CHARGE syndrome patients - correlation with audiological results and proposal for computed tomography scans evaluation methodology. Brazilian Journal of Otorhinolaryngology. 2024;90(2):101383. DOI: 10.1016/j.bjorl.2023.101383.

Jednym z głównych kryteriów rozpoznania zespołu CHARGE, będącego zespołem wad zrodzonych, są anomalie ucha. W trzecim artykule, stanowiącym część cyklu rozprawy doktorskiej, szczegółowo opisano spektrum wad wrodzonych ucha wewnętrznego u pacjentów z zespołem CHARGE, stosując ten sam protokół rekonstrukcji obrazów CT jak w pierwszym i

drugim artykule cyklu. Grupę badaną stanowiło 10 pacjentów (20 uszu), a analizie poddano nie tylko badania obrazowe, lecz również wyniki badań audiologicznych. Szczególną uwagę zwrócono na wizualizację w postaci rycin i opis malformacji dotyczących ślimaka oraz przedsionka i kanałów półkolistych, z uwagi na ich charakterystyczne i częste występowanie w kościach skroniowych pacjentów z zespołem CHARGE. Dzięki temu kompleksowemu podejściu możliwe jest lepsze zrozumienie wad rozwojowych ucha wewnętrznego oraz ich wpływu na słuch, co jest kluczowe dla opracowania skutecznych strategii rehabilitacji audiologicznej.

Wnioski

Przedstawione publikacje stanowiące cykl rozprawy doktorskiej wnoszą wkład w dziedzinie audiologii i otologii do diagnostyki i leczenia niedosłuchu poprzez nowatorskie opracowanie i szczegółowe przedstawienie protokołu metodologii rekonstrukcji i analizy obrazów tomografii komputerowej oraz jego zastosowania do oceny wad wrodzonych ucha wewnętrznego w praktyce klinicznej. Przedstawiona jest również korelacja szczegółów anatomicznych malformacji ucha wewnętrznego z wynikami audiologicznymi.

Wiedza uzyskana z tak opracowanych obrazów TK kości skroniowych jest istotna z praktycznego punktu widzenia i może być bardzo przydatna do dalszych badań i opracowywania bardziej precyzyjnych i zindywidualizowanych metod leczenia niedosłuchu w grupie pacjentów z wadami wrodzonymi ucha wewnętrznego.

Streszczenie w języku angielskim

Introduction

Congenital anomalies of the inner ear, originating during embryogenesis, can be visualized through imaging studies. These pathologies account for approximately 20% of congenital sensorineural hearing loss cases and are characterized by significant anatomical variation and a broad spectrum of clinical, including audiological, severity. Advancements in technology have facilitated increasingly precise imaging through high-resolution computed tomography and magnetic resonance imaging, leading to the development of several classifications used in clinical practice. The most comprehensive among these is the Sennaroğlu classification, initially introduced in 2002 and updated in 2017 to incorporate recent findings.

Identifying congenital inner ear defects is crucial for customizing suitable hearing rehabilitation approaches. Some defects can be corrected with hearing aids, while others, such as profound bilateral sensorineural hearing loss, require cochlear implantation.

Aims

1. Development of a methodology for multiplanar reconstruction of computed tomography images of the temporal bones of patients with inner ear congenital defects, supplemented with 3D reconstruction.
2. Application of the developed protocol for reconstruction and analysis of inner ear computed tomography images in clinical practice in counseling patients with inner ear defects for cochlear implantation.
3. Using a developed protocol, assessment of congenital defects of the inner ear in patients with CHARGE syndrome and correlation of reconstructed computed tomography images of the inner ear with the results of audiological tests.

Manuscript #1

Szleper A, Lachowska M, Pastuszka A, Łukaszewicz-Moszyńska Z, Wojciechowski T, Niemczyk K. Anatomical and clinical aspects and outcomes of bilateral cochlear implantation in cochlear hypoplasia type IV – a case report. Polski Przegląd Otorynolaryngologiczny. (2022);11(4):57-63. DOI: 10.5604/01.3001.0016.2238.

The first publication initiates a series of investigations on imaging studies, here extended to include auditory rehabilitation and its initial results. In the described case, a 6-month-old patient was diagnosed with a bilateral congenital inner ear malformation—type IV cochlear

hypoplasia. Multiplanar reconstruction of the computed tomography images and 3D reconstruction were performed to conduct a detailed analysis of the middle and inner ear anatomy. Both modalities of imaging reconstruction played an important role in determining appropriate electrode selection for cochlear implantation.

Manuscript #2

Szleper A, Lachowska M, Wojciechowski T, Niemczyk K. Computed tomography multiplanar and 3D image assessment protocol for detailed analysis of inner ear malformations in patients undergoing cochlear implantation counseling. Otolaryngologia Polska. 2024;78(2):35-43. DOI: 10.5604/01.3001.0054.2567.

The article presents a protocol for multiplanar reconstruction of inner ear CT images supplemented with 3D reconstructions, which helps clinicians better understand the defect's nature and facilitate therapeutic decision-making. The second manuscript is a detailed presentation of the CT scan analysis protocol used in the first article of the series. The analyzed group consisted of patients undergoing imaging and audiological diagnostics during qualification for cochlear implantation. Eleven patients (22 ears) with a detected congenital inner ear defect were analyzed retrospectively. The findings were presented in numerous, clear visualizations complemented by descriptive data, enabling a more precise presentation of the study outcomes.

Manuscript #3

Szleper A, Lachowska M, Wojciechowski T, Pronicka-Iwanicka K. Detailed analysis of inner ear malformations in CHARGE syndrome patients - correlation with audiological results and proposal for computed tomography scans evaluation methodology. Brazilian Journal of Otorhinolaryngology. 2024;90(2):101383. DOI: 10.1016/j.bjorl.2023.101383.

One of the main criteria for diagnosing CHARGE syndrome, a syndrome of congenital disabilities, is ear anomalies. In the third manuscript, the spectrum of inner ear congenital defects in patients with CHARGE syndrome is described in detail, using the same CT image reconstruction protocol as in the first and second articles of the series. The study group consisted of 10 patients (20 ears), and not only imaging tests but also the results of audiological tests were analyzed. Special attention was given to the visualization and the description of malformations affecting the cochlea, vestibule, and semicircular canals due to their typical and frequent manifestation in the temporal bones of patients with CHARGE syndrome. This comprehensive approach facilitates a better understanding of developmental inner ear abnormalities and their

impact on hearing function, which is crucial for developing effective strategies for audiological rehabilitation.

Conclusions

The presented manuscripts, constituting a series of doctoral dissertation, contribute to the field of audiology and otology in the diagnosis and treatment of hearing loss through the innovative development and detailed presentation of the protocol for the reconstruction and analysis of computed tomography images and its application to the assessment of congenital defects of the inner ear in clinical practice. The correlation between the anatomical details of inner ear malformations and audiological findings is also presented.

The knowledge obtained from such reconstruction of CT images of the temporal bones is important from a practical point of view. It may be very useful for further research and development of more precise and individualized methods of treating hearing loss in a group of patients with congenital defects of the inner ear.

Wady wrodzone ucha wewnętrznego - nowoczesna metoda obrazowania tomografią komputerową i jej znaczenie w audiologii

Wstęp

Pierwszy raport dotyczący wad wrodzonych ucha wewnętrznego został stworzony przez Carlo Mondiniego w 1791 r. Pomimo, iż opisana wówczas wada stanowi jedynie niewielki wycinek całości spektrum wad wrodzonych ucha, przez prawie dwa stulecia w odniesieniu do praktycznie każdej z nich używano nazwy deformacja Mondiniego lub dysplazja Mondiniego. Rozwój technik badań obrazowych przyniósł przełom również w diagnostyce zaburzeń rozwoju kości skroniowej. Klasyfikacja zaproponowana przez Leventa Sennaroglu w 2002 roku, kolejno modyfikowana aż do ostatecznej wersji z 2017 roku, jest obecnie najczęściej używaną na świecie. Do jej zalet należy przestrzeganie ugruntowanej nomenklatury i rozwinięcie o nowoczesne wyniki badań radiologicznych oraz możliwości rehabilitacji narządu słuchu. Klasyfikacja ta prezentuje wady od całkowitej aplazji błędnika, poprzez spektrum hipoplazji, aż do wad z zakresu niekompletnego podziału ślimaka.

Prezentowany cykl publikacji stanowiących rozprawę doktorską, wykorzystuje istniejącą najnowszą klasyfikację wad wrodzonych ucha wewnętrznego i proponuje nowe praktyczne podejście do metody analizy badań obrazowych oraz ich odniesienie do wyników audiologicznych. Grupy badawcze stanowią pacjenci, którzy byli poddawani diagnostyce w celu wyboru jak najlepszej i indywidualnie dostosowanej metody leczenia niedosłuchu.

W przypadku rozważania przeprowadzenia procedury wszczepienia implantu ślimakowego niezwykle istotna staje się znajomość anatomii ucha wewnętrznego kwalifikowanego pacjenta. Niektóre wady ucha wewnętrznego wykluczają bowiem możliwość przeprowadzenia tej procedury chirurgicznej pomimo spełniania kryteriów audiologicznych. Ponadto wady wrodzone ucha wewnętrznego każdorazowo stanowią wyzwanie dla chirurga wszczepiającego implant ślimakowy w związku z ryzykiem m.in. wystąpienia niekontrolowanego wypływu płynu mózgowo-rdzeniowego czy anomalii przebiegu nerwu twarzowego. Każda taka operacja powinna być poprzedzona szczegółową analizą obrazów TK kości skroniowych i właściwym wyborem dostępu chirurgicznego oraz rodzaju elektrody implantu ślimakowego.

Z uwagi na niskie rozpowszechnienie wad wrodzonych ucha wewnętrznego w populacji ogólnej, ukazujące się w literaturze medycznej publikacje charakteryzują się małolicznymi grupami badawczymi, zatem każda kolejna praca dotycząca tej tematyki stanowi istotne ugruntowanie i/lub uzupełnienie dotychczasowej wiedzy. Publikacje stanowiące cykl prezentowanej rozprawy doktorskiej, poza nowatorskim i praktycznym w audiologii i otologii wymiarem stosowania zaproponowanego w nich protokołu rekonstrukcji wielopłaszczyznowej i analizy obrazów tomografii komputerowej (CT), wykorzystują innowacyjne przedstawienie wad w postaci rekonstrukcji 3D. Zaproponowany protokół rekonstrukcji wraz ze szczegółowym opisem analizowanych obrazów CT sprawia, że istota malformacji ucha wewnętrznego staje się łatwiejsza do zrozumienia, a przedstawione wyniki badań audiologicznych prezentowanych pacjentów wykazują korelację ze stopniem zaburzenia rozwoju ucha wewnętrznego.

Szczególną uwagę skupiono na podgrupie, która wyróżniała się na tle całości pacjentów swoją złożonością i różnorodnością wad wrodzonych ucha wewnętrznego. Pacjenci z zespołem CHARGE, dla którego wady ucha stanowią jedno z dużych kryteriów rozpoznania zespołu, prezentują spektrum wad dotyczących zarówno ślimaka jak i przedsionka oraz kanałów półkolistych.

Pierwszą publikację prezentowanego cyklu rozprawy doktorskiej (*Szleper A, Lachowska M, Pastuszka A, Łukaszewicz-Moszyńska Z, Wojciechowski T, Niemczyk K. Anatomical and clinical aspects and outcomes of bilateral cochlear implantation in cochlear hypoplasia type IV – a case report. Polski Przegląd Otorynolaryngologiczny. (2022);11(4):57-63. DOI: 10.5604/01.3001.0016.2238)* stanowi opis przypadku wraz z przeglądem literatury. Opis dotyczy 6-miesięcznej pacjentki, u której w procesie diagnostycznym niedosłuchu wykryto wadę rozwojową ucha wewnętrznego – hipoplazję ślimaka typu IV. W celu przeprowadzenia szczegółowej analizy wspomnianej wady na podstawie badania CT kości skroniowych wykonano autorsko opracowaną rekonstrukcję wielopłaszczyznową obrazów CT uzupełnioną o rekonstrukcję 3D uwidaczniające szczegóły anatomiczne ucha wewnętrznego pacjentki, tj. zachowany zakręt podstawny ślimaka z hipoplastycznymi i przesuniętymi przednio i przyśrodkowo zakrętami środkowym i szczytowym. Ponadto stwierdzono brak kanałów półkolistych bocznych z rozwiniętymi prawidłowo kanałami półkolistymi przednim i tylnym. Uzyskane obrazy przedstawiono w formie szczegółowych rycin, a dla lepszego uwidocznienia i zrozumienia patologii oraz porównania zamieszczono uzyskane w tym samym protokole przykładowe obrazy prawidłowego ucha wewnętrznego. Badania audiologiczne przedstawianej pacjentki wykazały obustronnie głęboki niedosłuch odbiorczy. W

prezentowanym przypadku wykonano sekwencyjnie obustronną implantację ślimakową. Podczas operacji wystąpiło oczekiwane niekompletne umieszczenie elektrody w ślimaku z uwagi na jego zaburzoną morfologię. W pierwszym implancie w ślimaku umieszczono i aktywowano 10 elektrod, w drugim 12, co potwierdzono pomiarami impedancji. Jednak badanie telemetrii odpowiedzi neuronowej (NRT) nie wykazało żadnej odpowiedzi ani w analizie automatycznej, ani manualnej. Pierwsze powtarzalne reakcje słuchowe zaobserwowano po 2 miesiącach od aktywacji drugiego implantu. Powyższy przypadek wykazał, iż indywidualne podejście diagnostyczne oraz terapeutyczne wraz z dobrą współpracą rodziców z terapeutami i odpowiednio zdefiniowane cele rozwojowe u danego pacjenta są kluczem do zoptymalizowania efektów protezowania słuchu w niestandardowych przypadkach klinicznych.

Druga publikacja prezentowanego cyklu rozprawy doktorskiej (*Szleper A, Lachowska M, Wojciechowski T, Niemczyk K. Computed tomography multi-planar and 3D image assessment protocol for detailed analysis of inner ear malformations in patients undergoing cochlear implantation counseling. Otolaryngologia Polska. 2024;78(2):35-43. DOI: 10.5604/01.3001.0054.2567*) to artykuł oryginalny. Przedstawiono w nim autorsko opracowany protokół wielopłaszczyznowej rekonstrukcji obrazów tomografii komputerowej kości skroniowych oraz wykorzystania rekonstrukcji 3D w celu szczegółowej analizy wad rozwojowych ucha wewnętrznego u pacjentów kwalifikowanych do operacji wszczepienia implantu ślimakowego. W pracy analizie poddano grupę 11 pacjentów (22 uszu) z wadami ucha wewnętrznego przebadanych w ramach diagnostyki pod kątem wszczepienia implantu ślimakowego. U wszystkich powyższych pacjentów zastosowano ten sam protokół składający się z usystematyzowanych wielopłaszczyznowych rekonstrukcji CT i rekonstrukcji 3D ucha wewnętrznego oraz z diagnostyki audiologicznej. Ten sam protokół był zastosowany w publikacji pierwszej przedstawianego cyklu.

Wyniki rekonstrukcji obrazów CT przedstawiono w formie szczegółowych rycin. Aby zachować czytelność osobno przedstawiono malformacje ślimaka oraz wady towarzyszące dotyczące części przedsionkowej ucha wewnętrznego. Dołączone objaśnienia pisemne zgrupowane w tabeli wraz z wynikami badań audiologicznych pozwalają uzyskać pełny obraz i ułatwiają zrozumienie istoty poszczególnych wad ucha wewnętrznego.

Ponadto omówione zostały powikłania śródoperacyjne, które wystąpiły u analizowanych pacjentów poddanych operacji implantacji ślimakowej. Artykuł ten, z uwagi na szczegółowo przedstawiony w nim protokół rekonstrukcji i interpretacji obrazów tomografii

komputerowej, stanowi wartościowy wkład do diagnostyki i indywidualizowania podejścia do leczenia wrodzonego niedosłuchu u pacjentów z wadami ucha wewnętrznego.

Trzecia publikacja prezentowanego cyklu rozprawy doktorskiej *(Szleper A, Lachowska M, Wojciechowski T, Pronicka-Iwanicka K. Detailed analysis of inner ear malformations in CHARGE syndrome patients - correlation with audiological results and proposal for computed tomography scans evaluation methodology. Brazilian Journal of Otorhinolaryngology. 2024;90(2):101383. DOI: 10.1016/j.bjorl.2023.101383)* rozpatruje poruszane wcześniej kwestie wizualizacji wad wrodzonych ucha wewnętrznego w zawężonym do zespołu CHARGE kontekście. Złożoność i szerokie spektrum wad towarzyszących tej asocjacji oraz niska częstość jej występowania podnosi wartość prezentowanej publikacji, która objęła 10 pacjentów (20 uszu). W przypadku wad ślimaka uwidoczniono szerokie spektrum malformacji począwszy od aplazji ślimaka z poszerzonym przedsionkiem (CADV) oraz resztkowej otocysty, przez hipoplazję ślimaka typu II i III, aż do prawidłowo wykształconego ślimaka z towarzyszącymi izolowanymi wadami części przedsionkowej ucha wewnętrznego. Najczęstszą anomalią dotyczącą narządu przedsionkowego u analizowanych pacjentów z zespołem CHARGE, występującą w przedstawianej pracy w 100% przypadków, był brak bocznego kanału półkolistego, choć badanie wykazało, że kanały tylny i górny (w kolejno 95% i 65% przypadków) również nie występowały. Ponadto wykazano istotną korelację pomiędzy nasileniem deformacji przedsionka a średnicą przewodu słuchowego wewnętrznego (IAC) - im poważniejsza jest deformacja przedsionka, tym mniejsza średnica IAC. Dodatkowo nasilenie wady przedsionkowej istotnie korelowało z nasileniem hipoplazji zakrętu podstawnego ślimaka. Co więcej, korelacja wystąpiła w przypadku malformacji przedsionkowej i obecności lub braku wrzecionka, jego brak korelował z nasileniem wady przedsionka. Również wyniki badań audiologicznych korelowały znacząco z nasileniem stopnia malformacji ślimaka. Im bardziej nasilona wada, a co za tym idzie zaburzenie architektury wewnętrznej ślimaka, tym gorsze wyniki audiologiczne.

Z powodu towarzyszących obciążeń internistycznych pacjenci z zespołem CHARGE niejednokrotnie nie mogą być zakwalifikowani do leczenia chirurgicznego mającego na celu poprawienie słyszenia pomimo pozytywnej kwalifikacji w wymiarze otologicznym i audiologicznym. Wyniki przedstawionego badania potwierdzają, że anomalie ucha wewnętrznego są charakterystyczną cechą zespołu CHARGE. Ich obecność ma istotne implikacje kliniczne. Niedosłuch jest powszechny u tych pacjentów. Właściwe opisanie wad ucha wewnętrznego w tej grupie chorych w korelacji z wynikami badań audiologicznych, a w

konsekwencji oszacowanie powodzenia dostępnych metod leczenia niedosłuchu, pozawala na zindywidualizowane podejście i optymalizację terapii.

Założenia i cel cyklu publikacji stanowiących rozprawę doktorską

1. Opracowanie metodologii rekonstrukcji wielopłaszczyznowej obrazów tomografii komputerowej kości skroniowych pacjentów z wadami wrodzonymi ucha wewnętrznego uzupełnionej rekonstrukcją 3D .
2. Zastosowanie opracowanego protokołu rekonstrukcji i analizy obrazów tomografii komputerowej ucha wewnętrznego w praktyce klinicznej w procesie kwalifikacji do implantacji ślimakowej pacjentów z wadami ucha wewnętrznego.
3. Ocena, przy użyciu wypracowanego protokołu, wad wrodzonych ucha wewnętrznego u pacjentów z zespołem CHARGE oraz korelacja wyników analizy zrekonstruowanych obrazów tomografii komputerowej ucha wewnętrznego z wynikami badań audiologicznych.

Kopie opublikowanych prac

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CASE STUDY

Anatomical and clinical aspects and outcomes of bilateral cochlear implantation in cochlear hypoplasia type IV – a case report

Anatomiczne i kliniczne aspekty oraz wyniki obustronnej implantacji ślimakowej w hipoplazji ślimaka typu IV – studium przypadku

Authors' Contribution:
A – Study Design
B – Data Collection
C – Statistical Analysis
D – Manuscript Preparation
E – Literature Search

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ABSTRACT:

Cochlear hypoplasia is a congenital inner ear malformation (IEM) characterized by a reduced external cochlear dimension, usually accompanied by an abnormal internal architecture. Type IV cochlear hypoplasia is a cochlea with hypoplastic middle and apical turns. It may occur along with dislocation of the facial nerve, associated with semicircular abnormalities, less clearly marked promontory, or stapedial fixation. Such patients can present a broad spectrum of audiological test results, from sensorineural or mixed mild to profound hearing loss. The above anatomical changes may be responsible for intraoperative difficulties during cochlear implantation. In the studied case, a 6-month-old patient was diagnosed with an inner ear malformation – cochlear hypoplasia type IV on both sides. Computed tomography with multiplanar and 3D reconstruction was performed to analyze the middle and inner ear anatomy in detail. Both types of imaging reconstruction helped decide which cochlear implant electrode to choose. Perimodiolar-positioned cochlear implant electrode was found to be the most suitable choice. The patient underwent sequential bilateral cochlear implantation with expected incomplete electrode array insertion on both sides. First repeatable auditory responses were observed 2 months after the second implant activation. Good parental cooperation with therapists and adequately defined developmental goals in the presented patient allowed the multidisciplinary team to take advantage of the child's intellectual abilities and choose a suitable communication method; however, the patient's auditory responses were obtained slowly. The final auditory results cannot be predicted in inner ear malformations due to abnormal anatomical structure and, thus, heterogeneous innervation within the deformed cochlea. The programming of the sound processor must be individual in each case, based on the child's behavior observation and, if possible, objective test results. Patients with cochlear malformations usually require higher stimulation intensities to obtain sound sensations than patients with a typical cochlear structure.

KEYWORDS: cochlear hypoplasia, cochlear implantation, congenital malformations, hearing loss, inner ear, speech therapy

STRESZCZENIE:

Hipoplazja ślimaka to wrodzona wada rozwojowa ucha wewnętrznego charakteryzująca się zmniejszonym zewnętrznym wymiarem ślimaka, któremu zwykle towarzyszy jego nieprawidłowa architektura wewnętrzna. Hipoplazja typu IV zdefiniowana jest jako ślimak z hipoplastycznym środkowym oraz szczytowym zakrętem. Może jej towarzyszyć zaburzony przebieg nerwu twarzowego, nieprawidłowości budowy kanałów półkolistych, mniej wyraźnie zaznaczone promontorium lub fiksacja strzemiączka. Pacjenci z taką patologią mogą prezentować szerokie spektrum wyników badań audiologicznych, od niedosłuchu odbiorczego do mieszanego oraz stopnia od niewielkiego do głębokiego. Powyższe zmiany anatomiczne mogą być odpowiedzialne za trudności śródoperacyjne podczas wszczepienia implantu ślimakowego w przypadku niedosłuchu głebokiego. W opisywanym przypadku u 6-miesięcznej pacjentki rozpoznano wadę rozwojową ucha wewnętrznego – obustronną hipoplazję ślimaka typu IV. Tomografia komputerowa z rekonstrukcją wielopłaszczyznową oraz 3D została wykorzystana w celu szczegółowej analizy anatomii ucha środkowego i wewnętrznego. Oba rodzaje rekonstrukcji obrazowej były pomocne w podjęciu decyzji o wyborze elektrody implantu ślimakowego. Elektroda perimodiolarna została uznana za najodpowiedniejszą w powyższym przypadku. Pacjentka została poddana sekwencyjnemu, obustronnemu wszczepieniu implantu ślimakowego ze spodziewanym, niepełnym wprowadzeniem elektrody do ślimaka po obu stronach. Pierwsze powtarzalne reakcje słuchowe zaobserwowano po 2 miesiącach od aktywacji drugiego implantu. Dobra współpraca rodziców z terapeutami i odpowiednio określone cele rozwojowe

w przedstawionym przypadku pozwoliły multidyscyplinarnemu zespołowi na wykorzystanie możliwości intelektualnych dziecka i dokonanie wyboru odpowiedniej metody komunikacji, jednakże odpowiedzi słuchowe u pacjentki uzyskiwano powoli. Ostateczne audiologiczne rezultaty wszczepienia implantów ślimakowych są trudne do przewidzenia w przypadku wad rozwojowych ucha wewnętrznego ze względu na nieprawidłową budowę anatomiczną, a zatem niejednorodność unerwienia w zdeformowanym ślimaku. Programowanie procesora dźwięku musi być przeprowadzane indywidualnie w każdym przypadku oraz oparte na obserwacji zachowania dziecka i, jeśli to możliwe, na obiektywnych wynikach badań. Pacjenci z wadami rozwojowymi ślimaka wymagają większej intensywności stymulacji w celu uzyskania wrażeń dźwiękowych niż pacjenci z typową budową ślimaka.

SŁOWA KLUCZOWE: hipoplazja ślimaka, implantacja ślimakowa, niedosłuch, terapia słuchowo-językowa, ucho wewnętrzne, wady wrodzone

ABBREVIATIONS

IEM – inner ear malformation
CH – cochlear hypoplasia
MPR – MultiPlanar Reconstruction

INTRODUCTION

Cochlear hypoplasia is the congenital inner ear malformation (IEM) characterized by a reduced external cochlear dimension, usually accompanied by an abnormal internal architecture. Type IV cochlear hypoplasia is a cochlea with hypoplastic middle and apical turns. It may occur along with dislocation of the facial nerve, associated with semicircular abnormalities, less clearly marked promontory, or stapedial fixation [1, 2]. Such patients can present a broad spectrum of audiological test results, from sensorineural or mixed mild to profound hearing loss. The above anatomical changes may be responsible for intraoperative difficulties during cochlear implantation.

CASE PRESENTATION

Patient description and clinical aspects

A 6-month-old girl was admitted to the department of otorhinolaryngology to extend the hearing loss diagnosis found in hearing screening tests after birth. The family history of hearing loss was negative. The patient was born at 41 weeks gestation by cesarean section with a score of 10 on the Apgar scale. In general pediatric assessment, a set of congenital defects was found, including narrowing of the branches of pulmonary arteries, umbilical hernia, skin aplasia in the right knee area, decreased muscle tone, and laryngomalacia. Otoscopy revealed a normal tympanic membrane, preceded by a wide, typical external auditory canal. An ABR showed bilateral profound sensorineural hearing loss accompanied by type A tympanogram and absent stapedius muscle reflexes on both sides. The patient started using hearing aids bilaterally at the age of 4 months.

Anatomical aspects of cochlear malformation

Computed tomography of the temporal bones was ordered to complement the diagnostics. It revealed bilateral cochlear malformations with preserved basilar turn and hypoplastic, anteriorly and medially located middle and apical turns (Fig. 1.). The deformity was classified as type IV cochlear hypoplasia [2]. In addition, there were no lateral semicircular canals on either side, with fully developed anterior and posterior semicircular canals (Fig. 2.). To accurately assess the inner ear's anatomy, we used the MultiPlanar Reconstruction (MPR) option to inspect the malformation details using 2D presentations (RadiAnt DICOM Viewer 2022.1 64-bit Medixant, Poznan, Poland). In addition, complementing the computed tomography, we performed 3D reconstruction in the Mimics Innovation Suite 24.0 program (Materialise, Belgium). Fig. 3., 4. show 2D presentations and 3D reconstruction of a normal ear to compare it to the congenital inner ear anomaly shown in Fig. 1., 2. for easier visual pathology identification.

In addition, referring to the work of Pamuk et al. [3], specific measurements of the malformed cochlea in the presented case were performed. In the described case, 3 measurements were possible: basal turn length, basal turn maximum height, and mean cochlear duct lateral wall length. The remaining measurements, cochlear canal midscalar and lateral wall length were impossible to perform due to the disturbed internal architecture of the cochlea and the lack of certainty as to the quality of their performance. The measurement results are presented in Tab. I. The CT scans with measurements of the right cochlea in the presented case are shown in Fig. 5. for a better understanding of methodology.

In the presented patient, both types of imaging reconstruction helped decide which cochlear implant electrode to choose. Perimodiolar-positioned cochlear implant electrode was found to be the most suitable choice. Cochlear sequential implantations on both sides were performed 10 months apart, with the first surgery taking place at the age of 10 months. Electrode placement in the malformed cochlea was possible through standard surgical access using posterior tympanotomy. On the right side, the electrode was inserted through cochleostomy, and on the left side, through the round window. The maximum electrode insertion was up to 10 electrode contacts on both sides, as predicted preoperatively. The postoperative period was uneventful, and the patient was dismissed for further rehabilitation.

Logopedic and psychological aspects

At first counseling for the first implant, the patient did not present any auditory reactions or pre-communication behaviors. She did not maintain eye contact, did not share the field of attention, and did not reach for objects with her hands. Frequent "freezing" moments in space were observed, and active communication with the child was impossible. For 6 months after the first cochlear implantation, no changes in the behavior or auditory progress of the child were observed. However, after 6 months, the first auditory responses appeared. The girl began to pay attention to the sounds of musical instruments, although these reactions were not stabilized. 1 year

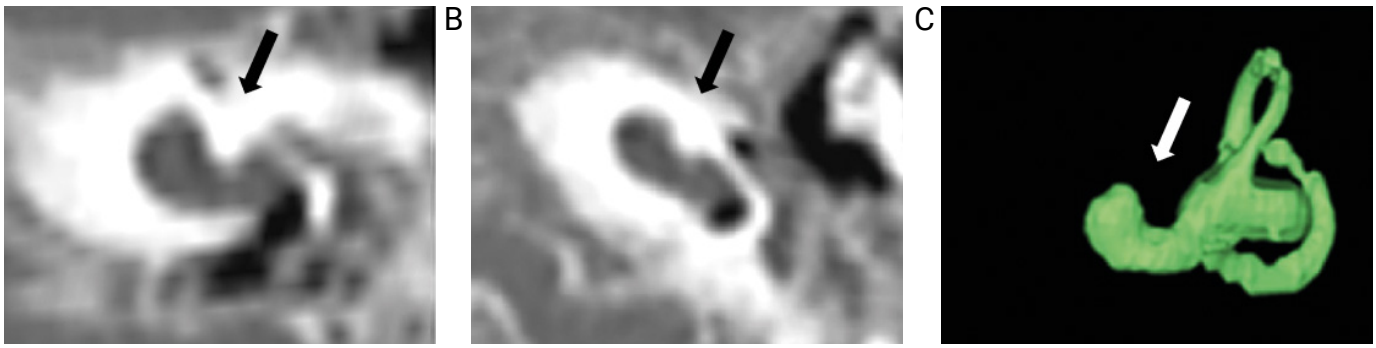

Fig. 1. Malformation of the cochlea in the presented case, shown in the planes selected for the most precise visualization in 2D presentation (A, B) and its (C) 3D reconstruction. Preserved basilar turn and hypoplastic, anteriorly and medially located middle and apical turns are indicated with arrows–cochlear hypoplasia type IV.

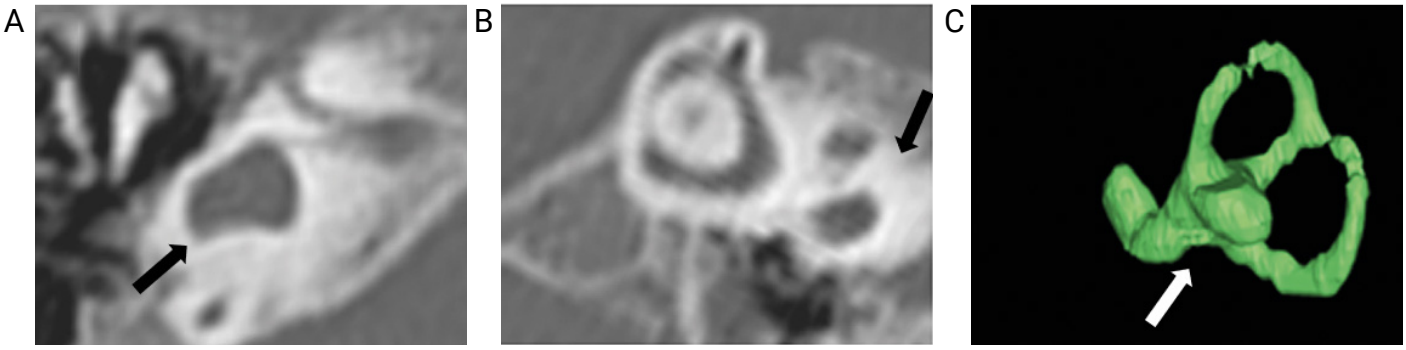

Fig. 2. Malformation of the semicircular canal in the presented case, shown in dedicated planes in 2D presentation (A, B) and its (C) 3D reconstruction. The absent lateral semicircular canal is indicated with arrows. Anterior and posterior semicircular canals are fully developed.

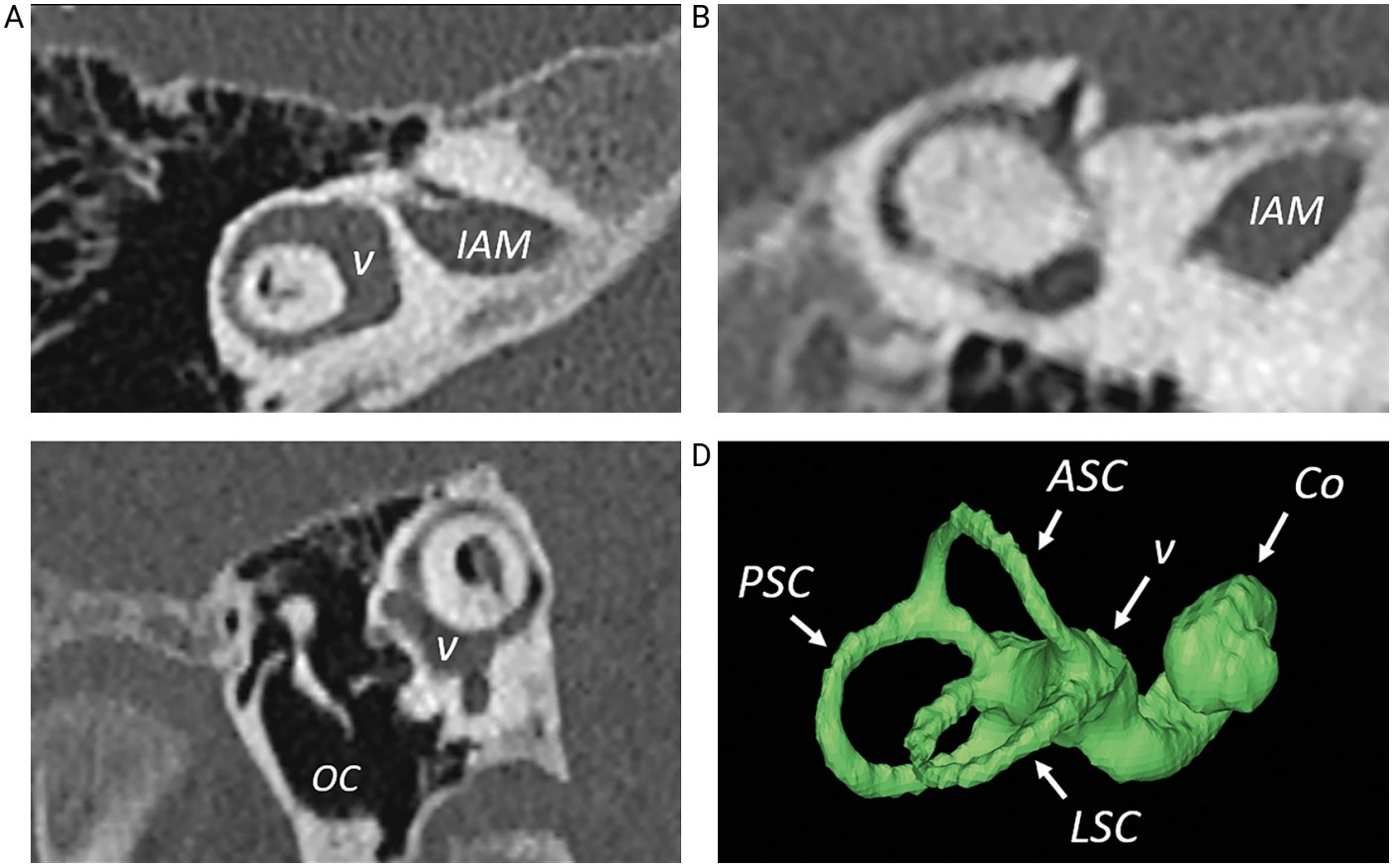

Fig. 3. Normal ear as reference shown with a set of multiplanar images of normal vestibular labyrinth presenting semicircular canals in the 3 reference planes (A, B, C) and its (D) 3D reconstruction. (A) shows the lateral semicircular canal in the axial plane, revealing a typical "signet ring appearance". (B) shows the plane of the anterior semicircular canal (as described by Pöschl). (C) shows the plane of the posterior semicircular canal with visible common crus. (D) shows 3D reconstruction. The markings in the panels present as follows: V – vestibule, IAM – internal acoustic meatus, OC – ossicular chain, PSC – posterior semicircular canal, ASC – anterior semicircular canal, LSC – lateral semicircular canal, Co – cochlea.

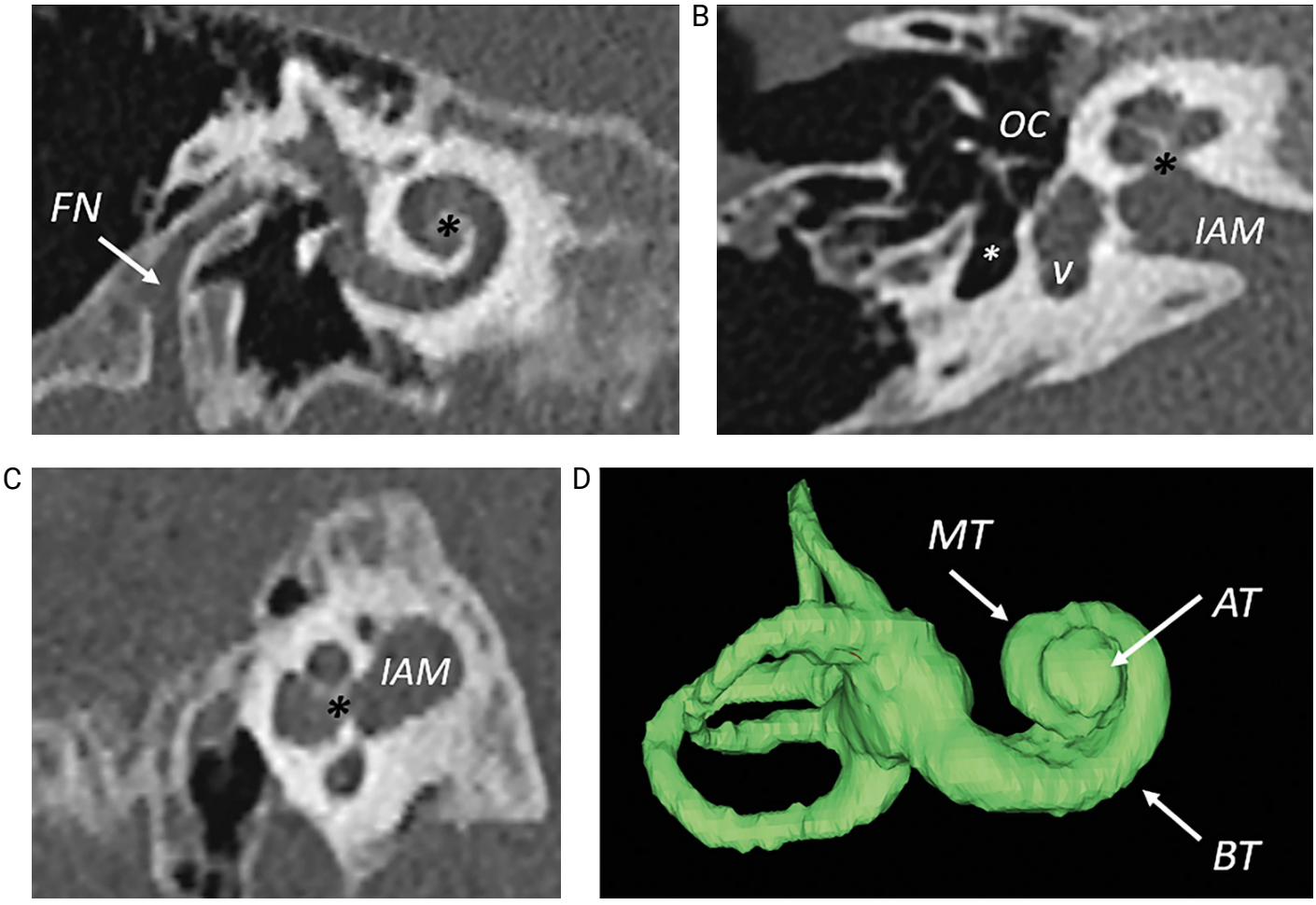

Fig. 4. Normal ear as reference shown with a set of multiplanar images of the normal cochlea and its 3D reconstruction. (A) shows the longitudinal section of the cochlea – the basal, middle, and apical turns around the modiolus (marked with a black "asterisk"). (B) shows the modified axial plane – the modiolus (marked with a black "asterisk") and all 3 cochlea turns; the tympanic cavity is marked with a white "asterisk". (C) shows the perpendicular plane to the modified axial-modiolus (marked with a black "asterisk") and all 3 turns of the cochlea. (D) shows 3D reconstruction. The markings in the panels present as follows: FN – facial nerve, BT – basal turn, MT – meddle turn, AT – apical turn.

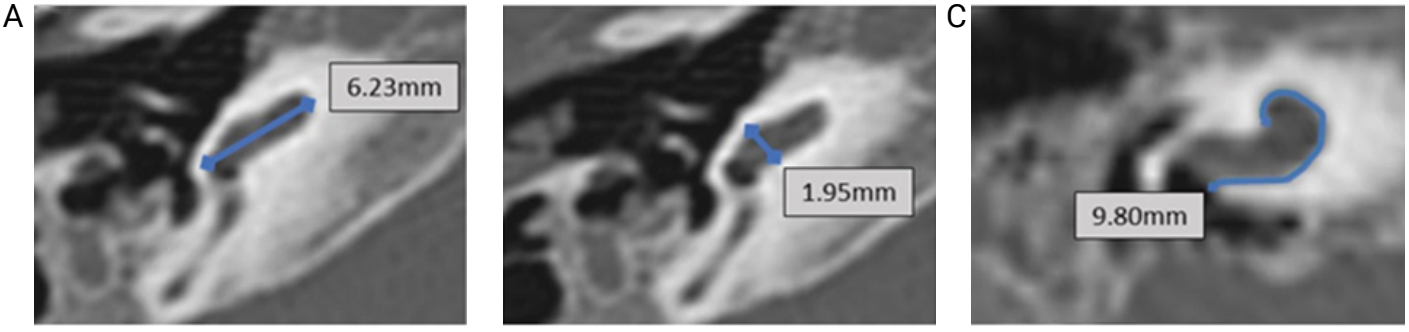

Fig. 5. Computed tomography axial reformatted images showing measurements (mm) of the right cochlea as described by Pamuk et al. [3]. (A) presents the measurement of the basal turn length (6.23 mm), (B) the basal turn maximum height (1.95 mm), and (C) the mean cochlear duct lateral wall length (9.80 mm).

after the first surgery, the first pre-communication behaviors were recorded. The child began to accost the people in the environment with her voice and pulled out her hand for toys.

1 year after the implantation of the first implant, the parents came to the department for second implant counseling for their daughter. During the process, information was provided about the potentially small benefits resulting from the use of bilateral implantation treatment. Due to the significantly delayed overall development of the child, no significant progress was expected in verbal communication. The parents continued her auditory education and logopedic rehabilitation. Due to the lack of progress in rehabilitation using the auditory-verbal method, the total communication method was introduced, with particular attention paid to gesture and sign language. Total communication focused on finding and using suitable communication methods in this case. It aimed to help the patient form connections and interactions with parents and therapists and, with time, ensure information exchange.

Tab. I. Results of the performed measurements of the malformed cochlea in the presented case as described by Pamuk et al. [3].

	Basal turn length (mm)	Basal turn maximum height (mm)	Cochlear duct outer wall mean length (mm)
Right cochlea	6.23	1.95	9.80
Left cochlea	5.62	2.01	10.2

After 5 months, the girl with 2 implants constantly reacted to all sounds of musical instruments. A labile but frequent reaction to her spoken name appeared. She actively communicated with her mother by pointing her finger at objects, perfectly reading the emotional state, and enforcing her needs by crying. When asked, the patient understood and pointed to several parts of the body and animals (onomatopoeia) when their sound was mimicked. At that point, several words in active speech and the passive dictionary appeared.

Postoperative processor fitting strategies

During the activation of the first implant in the right ear, 10 electrodes (el 13–22, the ones that were inserted during surgery) were activated, and the impedances of electrodes were correct. The neural response telemetry (NRT) measurements showed the patient's lack of reaction during the measurements and no neural response in automatic and manual analysis. The patient did not respond behaviorally to thresholds' determination for effective stimulation. 4 programs were written in the processor, varying in intensity – the parents were recommended to observe the child's reaction. During subsequent follow-up visits (1, 2, 3, 4, and 6 months after the activation), the intensities of electric stimulation were gradually increased – without unambiguous responses to the stimulation of individual electrodes and in "live mode" to any sounds up to 8 months after activation.

12 months after activating the first implant (right ear), the implant in the second ear (left) was activated. The parameters during activation and the procedure were analogous to the implant in the right ear. Adjustments were made based on the manually determined thresholds of the auditory nerve response; the records from individual electrodes were differentiated in terms of potentially present neural responses – electrodes that differed in recording (no response versus potentially present responses) were distinguished. On this basis, 12 consecutive electrodes were distinguished (11–22). The approximate settings of the C threshold below the NRT threshold were inspected for the child's response – both to the activation/deactivation of stimulation and the observation of the response to sounds. No reactions were observed at the moment; however, carefully observing the child's reactions was recommended to the parents. At subsequent follow-up visits, NRT inconclusive responses to stimulation were observed. After 2 months, an attempt to determine the thresholds was repeated, and the thresholds were manually determined. 11 effective stimulating electrodes were obtained (no response from electrode 21 was observed; however, it was left in the active form). 2 months after the activation of the processor in the second ear (left), repeatable auditory responses were observed in the first implanted ear (right). Due to the progress in rehabilitation and the first successful designated indicative hearing threshold determination by play audiometry, along with the help of assessment of the hearing threshold by a speech therapist, the thresholds of high-frequency stimulation were raised. However, careful attention was paid not to exceed the comfortable hearing thresholds. Further rehabilitation and observation of the child's reaction to sounds were recommended.

DISCUSSION

Cochlear hypoplasia (CH) was fully described for the first time by Sennaroglu et al. [1], who distinguished 4 types of this anomaly. Apart from the anomalies described in the presented case, this defect may be accompanied by an aberrant course of the tympanic segment of the facial nerve requiring modification of the surgical technique or intraoperative facial nerve monitoring. Moreover, stapes fixation may also be present in patients with CH types III and IV, and these patients may benefit from stapedotomy [2]. In addition, malformations of the semicircular canals may be observed. Li et al. [4] described the case of a patient with CH type IV with a sigmoid sinus obscuring the facial recess such that a posterior tympanotomy or retro-facial approach was impossible to perform, forcing a trans-meatal approach assisted by an endoscope. In our patient's CT scans, the course of the facial nerve and sigmoid sinus location seemed normal (typical), which did not require any modification of the surgical approach to the middle. An approach through atromastoidectomy and posterior tympanotomy was performed. However, the absence of the lateral semicircular canal, which serves as an easy referring point approaching the middle ear safely, made it a little more difficult to identify the position of the facial nerve during surgery and perform posterior tympanotomy. For an experienced otosurgeon, this small difficulty is quite easy to overcome, as it was in our case. Eventually, the surgery did not cause any complications.

In the study by Pamuk et al. [3], morphometric measurements carried out among cochleas with type IV hypoplasia showed no significant differences in basal turn length between CH type IV patients and the control normal group (unlike in CH types I–III). However, the basal turn maximum height differed significantly when comparing each type of hypoplastic cochleas to the normal group. Moreover, the mean mid-modiolar height was significantly different between those with CH type I and those with types II–IV. The measurements in our patient were partly consistent with Pamuk's results. Basal turn length and maximum height were within the ranges for type IV hypoplasia. However, the mean cochlear duct lateral wall length was significantly smaller and corresponded to the ranges found in type I hypoplasia. More research is needed on how to measure in detail the curve in the case of malformations with a more difficult or unclear internal cochlear architecture.

Incomplete cochlear implant electrode insertion in patients with inner ear malformations is reported in the literature more often than in patients without one. In Farhood et al. [5] study, complete insertion was seen in 81.8% of all IEMs compared with non-IEM cohorts, which achieved 98%. In Melo et al. [5] study, incomplete electrode

insertion was necessary for 3.8% of patients with cochlear hypoplasia. In Adunka et al. [6] and Isaiah et al. [7] studies, 4.9% and 6.4% of patients with IEMs had incomplete electrode insertion, respectively. In our case, incomplete electrode insertion was an expected complication during electrode selection before surgery. The choice in our case was the perimodiolar cochlear implant electrode, which, due to its construction idea, adapts to the shape of the modiolus, and thus is positioned closer to it, providing more focused stimulation with a lower current level, which results in more focused neural stimulation and lowers current consumption [8–10]. The cochlear hypoplasia type IV mainly involves the absence of the middle and apical turns, with relatively well-preserved basal turn. The choice of the perimodiolar electrode to fully exploit basal turn potential seemed reasonable. As mentioned before, the problem of incomplete electrode array insertion was expected. Above all, it was easy to overcome by activating only a certain number of contacts inserted in the cochlea. The electrodes left outside were not activated, which is fully reasonable.

In the statistical analysis demonstrated by Melo et al. [5] and Bille et al. [11], the children without inner ear malformations did not achieve statistically significantly better scores than children with one. However, the researchers noted that there were no children in the study group with significant comorbidities or mental retardation, which is considered to negatively influence the outcome of cochlear implantation. Black et al. [12] pointed out 2 main prognostic factors in the systematic literature review: medical/surgical and speech/language. In the first category, negative prognostic factors include, among others, severe malformations of the inner ear, which include hypoplasia and aplasia. An isolated enlarged vestibular aqueduct is considered the best prognosis, while cochlear nerve hypoplasia presents the worst prognosis of all IEMs [7]. In the second group, amongst prognostic factors, the cognitive delay, especially in the coincidence with the motor delay, seems to cause slower speech perception development skills [12–14]. In the presented case, apart from the developmental defect of the cochlea, the main obstacle in achieving satisfactory results of implantation seemed to be a delay in cognitive development. The absence of eye contact and visual interest in the environment, difficulties in sharing the field of attention, "freezing" moments in space, and slower overall perception were observed. These noticeably changed after rehabilitation with cochlear implants; however, the child's development still deviated from the age-appropriate norm.

The final auditory results cannot be predicted in inner ear malformations due to abnormal anatomical structure and, thus, heterogeneous innervation within the deformed cochlea. The programming of the sound processor must be individual in each case, based on the child's behavior observation and, if possible, objective test results, i.e., measurements of electrically evoked potentials from the distal part of the auditory nerve [15]. Regardless of the possibility of objective evaluation of the stimulation thresholds or their absence, the patient's behavior assessment should be used to determine the effective stimulation thresholds; in such cases, the experience of the person programming the implant processor and assessing the child's reactions is extremely important [16, 17]. In our case, based on the observations of parents and speech therapists, the intensities of electric stimulation were gradually increased, especially the thresholds of high-frequency stimulation. Patients with cochlear malformations usually require higher stimulation intensities to obtain sound sensations than patients with a typical cochlear structure [18].

CONCLUSIONS

As shown in the presented case, patients with inner ear malformations require a more individual approach to surgery and rehabilitation and higher stimulation intensities to obtain sound sensations. In the studied case, a 6-month-old patient was diagnosed with an inner ear malformation – cochlear hypoplasia type IV. CT with multiplanar and 3D reconstruction was performed to analyze the middle and inner ear anatomy in detail. The patient underwent sequential bilateral cochlear implantation with expected incomplete electrode array insertion on both sides. 10 electrode contacts in the first implant and 12 in the second were activated, confirmed with impedance measurements. However, the neural response telemetry (NRT) showed no response either in automatic or manual analysis. First repeatable auditory responses were observed 2 months after the second implant activation. Good parental cooperation with therapists and adequately defined developmental goals in the presented patient allowed the multidisciplinary team to take advantage of the child's intellectual abilities and choose the suitable communication method; however, the patient's auditory responses were obtained slowly.

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INDEX COPERNICUS INTERNATIONAL

ORIGINAL ARTICLE

Computed tomography multi-planar and 3D image assessment protocol for detailed analysis of inner ear malformations in patients undergoing cochlear implantation counseling

Authors' Contribution:
A – Study Design
B – Data Collection
C – Statistical Analysis
D – Data Interpretation
E – Manuscript Preparation
F – Literature Search
G – Funds Collection

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ABSTRACT:

Introduction: Congenital inner ear malformations resulting from embryogenesis may be visualized in radiological scans. Many attempts have been made to describe and classify the defects of the inner ear based on anatomical and radiological findings.

Aim: The aim was to propose and discuss computed tomography multi-planar and 3D image assessment protocols for detailed analysis of inner ear malformations in patients undergoing cochlear implantation counseling.

Material and methods: A retrospective analysis of 22 malformed inner ears. CT scans were analyzed using the Multi-Planar Reconstruction (MPR) option and 3D reconstruction.

Results: The protocol of image interpretation was developed to allow reproducibility for evaluating each set of images. The following malformations were identified: common cavity, cochlear hypoplasia type II, III, and IV, incomplete partition type II and III, and various combinations of vestibule labyrinth malformations. All anomalies have been presented and highlighted in figures with appropriate descriptions for easier identification. Figures of normal inner ears were also included for comparison. 3D reconstructions for each malformation were presented, adding clinical value to the detailed analysis.

Conclusions: Properly analyzing CT scans in cochlear implantation counseling is a necessary and beneficial tool for appropriate candidate selection and preparation for surgery. As proposed in this study, the unified scans evaluation scheme simplifies the identification of malformations and reduces the risk of omitting particular anomalies. Multi-planar assessment of scans provides most of the necessary details. The 3D reconstruction technique is valuable in addition to diagnostics influencing the decision-making process. It can minimize the risk of misdiagnosis. Disclosure of the inner ear defect and its precise imaging provides detailed anatomical knowledge of each ear, enabling the selection of the appropriate cochlear implant electrode and the optimal surgical technique.

KEYWORDS: cochlear implantation, computed tomography, congenital abnormalities, inner ear

ABBREVIATIONS

ABR – auditory brainstem response
ASC – anterior semicircular canal
ASSR – auditory steady-state response
CSF – cerebrospinal fluid
CT – computed tomography
HU – Hounsfield Units
LSC – lateral semicircular canal
MPR – Multi-Planar Reconstruction
PSC – posterior semicircular canal

INTRODUCTION

Congenital inner ear malformations resulting from embryogenesis may be visualized in radiological scans. According to the literature, they account for approximately 20% of the cases of sensorineural hearing loss [1]. The above abnormalities may cover the entire spectrum of severity and clinical symptoms. Many attempts have been made to describe and classify the defects of the inner ear based on anatomical and radiological findings. Over the years, more precise computed tomography with high resolution and magnetic resonance imaging studies allowed several classifications

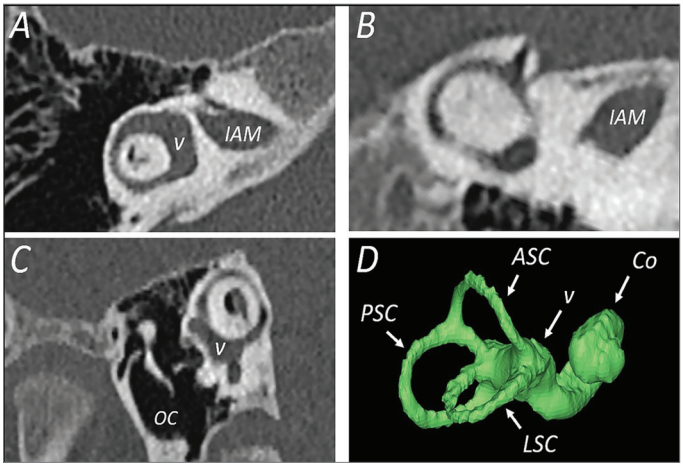

Fig. 1. Multi-planar images of a normal vestibular labyrinth presenting semicircular canals in the three reference planes and its 3D reconstruction. (A) the lateral semicircular canal presented in axial plane showing a typical "signet ring appearance"; (B) the plane of the anterior semicircular canal (as described by Pöschl); (C) the plane of the posterior semicircular canal with common crus; (D) 3D reconstruction. V – vestibule; IAM – internal acoustic meatus; OC – ossicular chain; PSC – posterior semicircular canal; ASC – anterior semicircular canal; LSC – lateral semicircular canal; Co – cochlea.

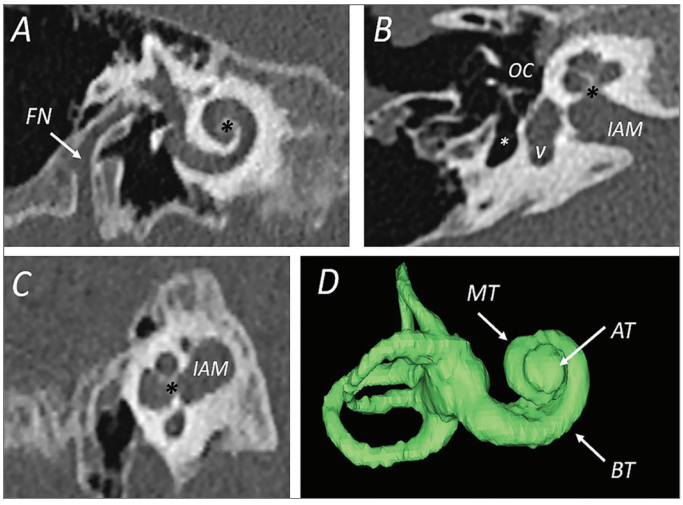

Fig. 2. Set of multi-planar images of the normal cochlea and its 3D reconstruction. (A) the longitudinal section of the cochlea – assessment of the basal, middle, and apical turns around the modiolus (marked with an 'asterisk'); (B) the modified axial plane – assessment of the modiolus and basal turn of the cochlea; (C) the perpendicular plane to the modified axial – the assessment of modiolus and all three turns of the cochlea; (D) 3D reconstruction. FN – facial nerve; BT – basal turn; MT – meddle turn; AT – apical turn.

to be used in clinical practice. The most fully describing the inner ear malformation is the Sennaroğlu classification, first drawn up in 2002, then modified to summarize the discoveries made so far, and finally adding a few innovations [2–5]. Some hearing defects can be corrected with hearing aids but most are profound bilateral sensorineural hearing impairments that indicate cochlear implantation.

The study aimed to take a closer and more detailed look at inner ear malformations in the light of imaging studies, point out potential challenges in classifying them, and develop new imaging modalities, such as 3D imaging, for the more informed otosurgeons performing cochlear implantation.

MATERIAL AND METHODS

Ethical consideration

This retrospective study was approved by the local Institutional Ethics Committee (decision number AKBE/175/2022). The project conforms to The Code of Ethics of the World Medical Association (Declaration of Helsinki).

Patients description

A total of 22 malformed inner ears in eleven patients were included in this retrospective study (11 consecutive patients with bilateral inner ear malformations). Ten patients were children aged between 7 months and 9 years old, and one was adult aged 33 years old. They were all referred to our Department due to cochlear implantation counseling and underwent the diagnostic procedure in the last five years (consecutive patients). This study analyzed the following data from medical records: medical and family history, audiological assessment, computed tomography (CT) scans, and surgical information (if, after the counseling, the patient was referred to the procedure).

Computed tomography scans analysis procedure

All the CT scans were obtained as a standard protocol in cochlear implantation patients' counseling. All scans were analyzed using RadiAnt DICOM Viewer 2022.1 64-bit (Medixant, Poznan, Poland) with Multi Planar Reconstruction (MPR) option, and 3D reconstruction images were obtained using Mimics Innovation Suite 24.0 (Materialise, Belgium).

The first step was to set the window level to 500 Hounsfield Units (HU) and the width to 3000 HU. Then, the protocol of image interpretation was developed to allow reproducibility for evaluating each set of images. The MPR tool was used to assess the semicircular canals, and the three reference planes were established. The axial plane was tilted, so the lateral semicircular canal (LSC) presented a typical "signet ring appearance". Subsequently, the plane of the anterior semicircular canal (ASC, as described by Pöschl) was found, and the plane of the posterior semicircular canal (PSC) with common crus was the last one to be found (Fig. 1.).

The next step was to describe in detail the cochlea and the vestibule. To fully visualize the cochlea, we proposed to assess it in three ways (Fig. 2.). At first, the longitudinal section of the cochlea was set to assess the basal, middle, and apical turns around the modiolus (in Fig. 2., marked with an 'asterisk'). Next, the modified axial plane was established, tilted to a position where the modiolus and basal turn of the cochlea could be assessed. Then, a perpendicular plane to the modified axial was set where the modiolus and all three turns of the cochlea could be visualized. To fully understand the variety of developmental defects found in analyzed malformed ears, 3D reconstructions were made and evaluated.

After carefully analyzing all ears, each malformation was classified according to the newest Sennaroglu classification [4].

Tab. I. Patients' data concerning gender, age of the diagnosis, auditory diagnostic results, and hearing loss type.

CASE NUMBER		#1	#2	#3	#4	#5	#6	#7	#8	#9	#10	#11
Gender		F	F	M	F	M	F	F	M	F	F	F
Age of the diagnosis		7 months	9 months	11 months	9 years	7 years	6 months	6 years	11 months	12 months	33 years	8 months
Hearing threshold (right ear)	500 Hz	NR	NR	NR	50 dBHL	NR	NR	NR	NR	35 dBnHL	100 dBHL	90 dBnHL
	1000 Hz	NR	NR	NR	65 dBHL	NR	NR	NR	NR	50 dBnHL	105 dBHL	90 dBnHL
	2000 Hz	NR	NR	NR	105 dBHL	NR	NR	NR	NR	45 dBnHL	NR	100 dBnHL
	4000 Hz	NR	NR	NR	NR	NR	NR	NR	NR	45 dBnHL	NR	100 dBnHL
Hearing threshold (left ear)	500 Hz	NR	NR	NR	55 dBHL	NR	NR	NR	NR	75 dBnHL	NR	>90 dBnHL
	1000 Hz	NR	NR	NR	55 dBHL	NR	NR	NR	NR	90 dBnHL	NR	90 dBnHL
	2000 Hz	NR	NR	NR	95 dBHL	NR	NR	NR	NR	90 dBnHL	NR	75 dBnHL
	4000 Hz	NR	NR	NR	NR	NR	NR	NR	NR	90 dBnHL	NR	55 dBnHL
Hearing loss type		SNHL	SNHL	SNHL	SNHL	SNHL	SNHL	SNHL	SNHL	conductive right ear/ mixed left ear	SNHL	mixed both ears

SNHL – sensorineural hearing loss; F – female; M – male; NR – no response; HL – hearing level; nHL – normalized hearing level.

RESULTS

The average patient age in the group was 5.10 years (SD = 9.64; median 0.92), with eight females and three males. Ten patients were children, and one was an adult female. Patients' age of the hearing loss diagnosis ranged from 6 months to 33 years old. Due to age and the possibility of cooperation, various hearing tests were performed. In case #4 and case #10, pure tone audiometry was performed. The remaining cases underwent auditory brainstem response (ABR) and auditory steady-state response (ASSR). All of the cases were diagnosed with hearing loss. The patients' demographics and audiology data are presented in Tab. I.

The total number of malformed ears was 22. In each case, inner ear malformation was present in both ears. Among the anomalies, 1 case of a common cavity, 2 cases of cochlear hypoplasia type II, 5 of cochlear hypoplasia type III, 4 of cochlear hypoplasia type IV, 3 of incomplete partition type II, and 2 cases of incomplete partition type III were found (all details are presented in Tab. II.). The malformed semicircular canals and vestibule were observed too. The most frequent concomitant defects were lateral semicircular canal anomalies, from hypoplastic to absent (found in 16 ears), usually accompanied by other vestibular labyrinth abnormalities. Moreover, in 5 cases, isolated malformations of the vestibular labyrinth were found. In 2 cases, the only abnormality was the absence of the posterior semicircular canal.

Four patients underwent unilateral cochlear implantation, and one underwent bilateral sequential cochlear implantation with a one-year interval between the surgeries. Five cochlear implant electrode insertions were performed through the standard operating procedure with posterior tympanotomy, and one required a retrofacial approach due to difficulties visualizing the round window. In 5 cases, cochleostomy was performed. In 1 case, an electrode was inserted through the round window. Moreover, during the cochlear implantation procedure, the surgeon faced such difficulties as a gusher or incomplete electrode insertions. A cerebrospinal fluid gusher was observed during surgery in

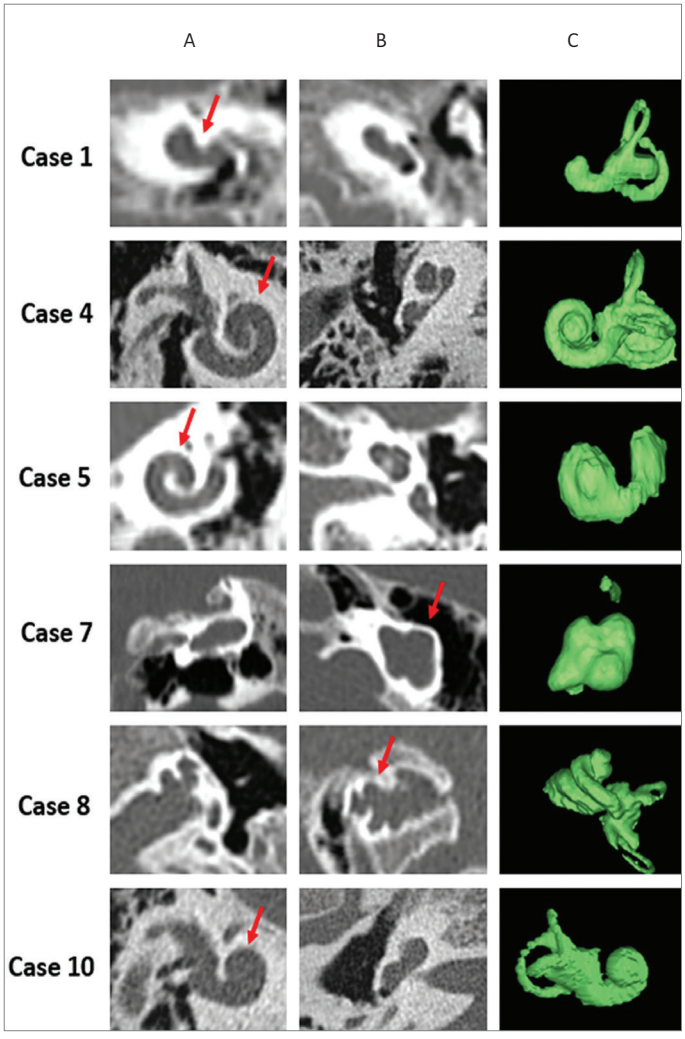

Fig. 3. Identified malformations of the cochlea presented in the planes selected for the most precise visualization (columns A and B) and its 3D reconstruction (column C). Each row represents a particular patient marked with an assigned number corresponding to numbers from Tab. I. and II. Red arrows indicate anomaly location: case #1 – cochlear hypoplasia type IV; case #4 – incomplete partition type II; case #5 – cochlear hypoplasia type III; case #7 – common cavity; case #8 – incomplete partition type III; case #10 – cochlear hypoplasia type II.

Tab. II. Computed tomography findings in the study group concerning vestibular labyrinth, cochlea, and other anatomical findings, along with the malformation classification according to Sennaroğlu [4].

CASE NUMBER	VESTIBULAR LABYRINTH	COCHLEA	OTHER FINDINGS (EVA/IAC)	CLASSIFICATION ACCORDING TO SENNAROĞLU (2017)
Case #1				
Right Ear	LSCC: absent ASCC: without pathology PSCC: without pathology	Basal turn preserved, middle and apical turns are severely hypoplastic and located anteriorly and medially, lack of internal structures: modiolus, interscalar septum		Cochlear hypoplasia type IV
Left Ear	LSCC: absent ASCC: without pathology PSCC: without pathology	Basal turn preserved, middle and apical turns are severely hypoplastic and located anteriorly and medially, lack of internal structures: modiolus, interscalar septum		Cochlear hypoplasia type IV
Case #2				
Right Ear	LSCC: shorter, dilated ASCC: without pathology PSCC: without pathology	Basal turn preserved, hypoplastic middle and apical turns		Cochlear hypoplasia type IV
Left Ear	LSCC: shorter, dilated ASCC: without pathology PSCC: without pathology	Basal turn preserved, hypoplastic middle and apical turns		Cochlear hypoplasia type IV
Case #3				
Right Ear	LSCC: without pathology ASCC: without pathology PSCC: hypoplastic	Without pathology	Slightly widened internal acoustic canal	Isolated SCCs anomaly
Left Ear	LSCC: without pathology ASCC: without pathology PSCC: absent	Without pathology	Slightly widened internal acoustic canal	Isolated SCCs anomaly
Case #4				
Right Ear	LSCC: without pathology ASCC: without pathology PSCC: without pathology Vestibule: mildly enlarged	Basal turn preserved, absent ISS between plump middle and apical turns, smooth external contour between middle and apical turns (baseball cap cochlea) Deficient modiolus	EVA	Incomplete partition type II
Left Ear	LSCC: without pathology ASCC: without pathology PSCC: without pathology Vestibule: mildly enlarged	Basal turn preserved, absence of the ISS between plump middle and apical turns, smooth external contour between middle and apical turns (baseball cap cochlea) Deficient modiolus	EVA	Incomplete partition type II
Case #5				
Right Ear	LSCC: absent ASCC: absent PSCC: absent	The cochlea has fewer turns (less than two turns) with a short modiolus	Arcuate eminence present, facial nerve present	Cochlear hypoplasia type III
Left Ear	LSCC: absent ASCC: absent PSCC: absent	The cochlea has fewer turns (less than two turns) with a short modiolus	Arcuate eminence present, facial nerve present	Cochlear hypoplasia type III
Case #6				
Right Ear	LSCC: hypoplastic, sclerotic ASCC: hypoplastic PSCC: absent	Without pathology		Isolated SCCs anomaly
Left Ear	LSCC: hypoplastic, sclerotic ASCC: hypoplastic PSCC: absent	Without pathology		Isolated SCCs anomaly

Tab. II. cd. Computed tomography findings in the study group concerning vestibular labyrinth, cochlea, and other anatomical findings, along with the malformation classification according to Sennaroğlu [4].

CASE NUMBER	VESTIBULAR LABYRINTH	COCHLEA	OTHER FINDINGS (EVA/IAC)	CLASSIFICATION ACCORDING TO SENNAROĞLU (2017)
Case #7				
Right Ear	LSCC: absent ASCC: absent PSCC: absent	Basal turn preserved, deficient modiolus, absence of the ISS, apical and middle turns forming a cystic apex		Incomplete partition type II
Left Ear	LSCC: absent ASCC: residual PSCC: absent	Cochlea and vestibule combined into one structure	EVA	Common cavity
Case #8				
Right Ear	LSCC: without pathology ASCC: without pathology PSCC: without pathology	Corkscrew cochlea placed directly at the lateral end of IAC without cribriform plate; absence of modiolus	Dilated IAC Gusher presented intraoperatively	Incomplete partition type III
Left Ear	LSCC: without pathology ASCC: without pathology PSCC: without pathology	Corkscrew cochlea placed directly at the lateral end of IAC without cribriform plate; absence of modiolus	Dilated IAC	Incomplete partition type III
Case #9				
Right Ear	LSCC: absent ASCC: absent PSCC: absent Vestibule: hypoplastic	Without pathology		Isolated vestibular labyrinth anomaly
Left Ear	LSCC: absent ASCC: absent PSCC: absent Vestibule: hypoplastic	The cochlea has fewer turns (less than two turns) with a short modiolus		Cochlear hypoplasia type III
Case #10				
Right Ear	LSCC: hypoplastic ASCC: hypoplastic PSCC: hypoplastic Absent common crus	Cochlea with smaller dimensions, absent modiolus, and interscalar septum		Cochlear hypoplasia type II
Left Ear	LSCC: hypoplastic ASCC: hypoplastic PSCC: absent	Cochlea with smaller dimensions, absent modiolus, and interscalar septum		Cochlear hypoplasia type II
Case #11				
Right Ear	LSCC: absent ASCC: absent PSCC: absent Vestibule: hypoplastic	The cochlea has fewer turns (less than two turns) with a short modiolus		Cochlear hypoplasia type III
Left Ear	LSCC: absent ASCC: absent PSCC: absent Vestibule: hypoplastic	The cochlea has fewer turns (less than two turns) with a short modiolus		Cochlear hypoplasia type III

SCC – semicircular canal; LSCC – lateral semicircular canal; ASCC – anterior semicircular canal; PSCC – posterior semicircular canal; ISS – interscalar septum; IAC – internal acoustic canal; EVA – enlarged vestibular aqueduct

one patient with incomplete partition type III (case #8). In this case, in the cochleostomy aperture, the electrode was secured with fragments of the temporal muscle, and no further leakage of the CSF was observed. Incomplete insertion was performed in cochlear implantations in case #2 with bilateral cochlear hypoplasia type IV.

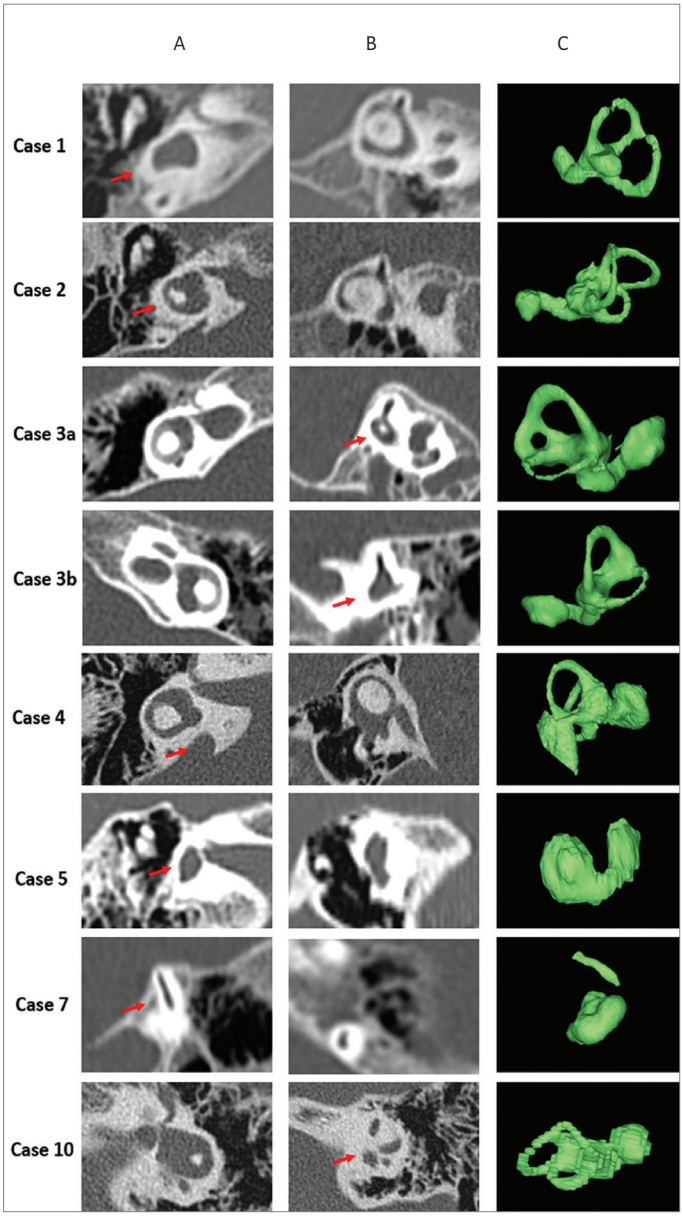

Fig. 4. Identified malformations of the semicircular canals presented in dedicated planes (columns A and B) and their 3D reconstruction (column C) – isolated defects of individual canals and complex defects. Each row represents a particular patient marked with an assigned number corresponding to numbers from Tab. I. and II. Red arrows indicate anomaly location: case #1 – absent LS; case #2 – hypoplastic LSC; case #3a right ear – hypoplastic PSC, and #3b left ear – absent PSC; case #10 – misplaced all three semicircular canals; case #5 – absent all semicircular canals; case #7 – residual, fragmentary ASC; case #4 – enlarged vestibular aqueduct.

Cochlear malformations were analyzed and compiled. The following malformations were found among our patients – common cavity (case #7 left ear), cochlear hypoplasia type II (case #10), cochlear hypoplasia type III (case #5, #9 left ear, and case #11), cochlear hypoplasia type IV (case #1 and #2), and incomplete partition type II (case #4 and case #7 right ear) and type III (case #8). The CT images have been compiled with 3D reconstructions to better visualize the pathology. The series of cochlear malformations found are presented in Fig. 3.

Subsequently, isolated and complex defects of individual canals were found in the spectrum of identified malformations of the semicircular canals. In Fig. 4., a series of isolated malformations found are presented: hypoplastic LSC (case #2), absent LSC (case #1), hypoplastic PSC (case #3 right ear), and absent PSC (case #3 left ear). More complex anomalies are presented, too. In case #10, the red arrow draws attention to the present but misplaced all three semicircular canals. In case #3, the absence of all semicircular canals is visualized, and in case #7, only residual and fragmentary ASC without any communication with the vestibule was identified. Case #4 presents the enlarged vestibular aqueduct. In one case (case #4), the 3D reconstruction cleared the doubts about identifying the malformation initially assigned as cochlear hypoplasia type III. The reconstruction highlighted the cystic apex of the cochlea, which resulted in the qualification change to incomplete partition type II.

DISCUSSION

For many years, it was believed that the inner ear's type of malformation depends on the time of the factor leading to the developmental arrest. The cochlear development is complete by the end of the 8th week. By the 11th week, the vestibule development is completed, and the semicircular canals by the end of the 22nd week [6].

The semicircular canals develop in the following order: anterior, posterior, and lateral. The absence of the posterior semicircular canal should also interfere with developing the lateral semicircular canal. Among the semicircular canals' malformations, four patients with the hypoplastic or absent posterior semicircular canal as the only abnormality were found in our study. Many genes responsible for complex malformations of the inner ear have been identified, including *Hoxa2, Hoxa1 / Hoxb1, kreisler,* and *Fgf3*. The *Netrin-1* gene mutation is believed to be responsible for all three SCC malformations, whereas *otx1, prx1/ prx2,* and *Nkx5-1* mutations lead mostly to malformations, including lateral SCC and lateral ampulla [7]. Further investigations in this area should be carried out.

Although the inner ear development occurs on a different embryological basis than the middle and external ear, combined malformations are found in 10% to 47% of cases [8]. Some of these abnormalities, such as the aplastic or obliterated round window or aberrant course of the facial nerve, may disturb classic surgical access to the round window for cochlear implantation, enforcing access modification, e.g. performing a retrofacial approach [9]. This modification was performed in one of our cochlear hypoplasia type IV patients. In middle and inner ear malformations, endoscopic assistance in surgery is becoming more popular [10, 11] but it was unnecessary in the described case #2.

According to the literature, gusher is the most common intraoperative complication in inner ear malformations. It occurs most often in patients with incomplete partition type III [12, 13], which is consistent with our observation. Moreover, according to Farhood et al. [14] systematic review, complete insertion was seen in 81.8% of all inner ear malformations, and CSF gusher was reported in 39.1% of cases.

Inner ear malformations are also comorbid in syndromic abnormalities, including CHARGE syndrome [15], where the

absence of semicircular canals is a frequently associated defect. Its prevalence can reach up to 100% [16, 17]. Among our group of patients, there were three children with CHARGE syndrome. In all of the cases, the defects identified in the study were cochlear hypoplasia type III (in 5 out of 6 ears), accompanied by the absence of all three semicircular canals on both sides.

CT is an examination dedicated to assessing the inner ear's bony structure, including diagnosing inner ear malformation. However, in our study, we observed several pitfalls that need to be considered in future studies. Various technicians' performance of computed tomography in different centers resulted in diversified examination parameters. In some cases, the quality of the performed scans was not entirely satisfactory concerning the possibility of assessing the bone structures of the inner ear. Given the above circumstances, it is necessary to standardize the examination's technical parameters and the method of tomography assessment. There are more and more radiological studies regarding congenital ear malformations and, therefore, more algorithms for their assessment [8, 15, 18, 19]. However, in clinical practice, we still observe isolated cases of incomplete radiological descriptions without the details necessary to correctly identify inner ear malformation. Therefore, a skilled and experienced radiologist and otosurgeon must assess imaging examinations of patients qualified for cochlear implantation. Our study proposes a method of assessing computed tomography and the planes where individual anatomical structures should be assessed.

In our study, it has been found that in individual cases, 3D reconstruction changed the assessment of the defect and, thus, the classification. Weiss et al. [20] already drew attention to the value of 3D reconstruction for correctly identifying the inner ear malformation in the differentiation between the common cavity and cochlear aplasia prior to cochlear implantation surgery. They reported a high rate of misdiagnosed cases of the common cavity. Nakamura et al. [21] faced another problem, and 3D segmentation was a helpful solution. In their case, a facial nerve anomaly in the form of nerve bifurcation did not allow cochlear implantation on the first attempt. Afterward, the 3D reconstruction resulted in a sufficiently clear anatomical image to plan the second surgery and perform the cochleostomy in this patient.

CONCLUSIONS

This study showed that the proper analysis of computed tomography scans in each cochlear implantation counseling is a necessary and beneficial tool for appropriate candidate selection and preparation for surgery. As proposed in this study, the unified scans evaluation scheme simplifies the identification of malformations and reduces the risk of omitting particular anomalies. Multi-planar assessment of computed tomography scans provides most of the necessary details and should be used by every otosurgeon performing cochlear implantations. A more advanced assessment technique, such as 3D reconstruction, is valuable in addition to diagnostics influencing the decision-making process. It can minimize the risk of misdiagnosis. Disclosure of the inner ear defect in the diagnostic process and its precise imaging provide detailed anatomical knowledge of each ear. It enables the selection of the appropriate cochlear implant electrode and the optimal surgical technique. The above aspects enable the maximum safety of cochlear implantation in patients with congenital inner ear malformations.

INFORMED CONSENT

This is a retrospective study, and informed consent was not required – this research study was conducted retrospectively from data obtained for clinical purposes.

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ORIGINAL ARTICLE

Detailed analysis of inner ear malformations in CHARGE syndrome patients – correlation with audiological results and proposal for computed tomography scans evaluation methodology

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Abstract
Objectives: The aim was to describe the spectrum of inner ear malformations in CHARGE syndrome and propose a Computed Tomography (CT) detailed scan evaluation methodology. The secondary aim was to correlate the CT findings with hearing thresholds.
Methods: Twenty ears of ten patients diagnosed with CHARGE syndrome were subjected to CT analysis focusing on the inner ear and internal acoustic canal. The protocol used is presented in detail. ASSR results were analyzed and correlated with inner ear malformations.
Results: Cochlear hypoplasia type III was the most common malformation found in 12 ears (60%). Cochlear hypoplasia type II, aplasia with a dilated vestibule, and rudimentary otocyst were also identified. In 20%, no cochlear anomaly was found. The lateral Semicircular Canal (SCC) absence affected 100% of ears, the absence of the posterior SCC 95%, and the superior SCC 65%. Better development of cochlea structures and IAC correlated significantly with the lower hearing thresholds.
Conclusion: This study demonstrated that rudimentary SCC or a complete absence of these SCCs was universally observed in all patients diagnosed with CHARGE syndrome. This finding supports the idea that inner ear anomalies are a hallmark feature of the CHARGE, contributing to its distinct clinical profile. The presence of inner ear malformations has substantial clinical implications. Audiological assessments are crucial for CHARGE syndrome, as hearing loss is common. Early detection of these malformations can guide appropriate interventions, such as

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hearing aids or cochlear implants, which may significantly improve developmental outcomes and communication for affected individuals. Recognizing inner ear malformations as a diagnostic criterion presents implications beyond clinical diagnosis. A better understanding of these malformations can advance the knowledge of CHARGE pathophysiology. It may also help guide future research into targeted therapies to mitigate the impact of inner ear anomalies on hearing and balance function.

Level of evidence: 4.

Introduction

CHARGE syndrome is a non-random congenital anomalies association that was first independently described in the medical literature in 1979 by Hall and Hittner,[1,2] then updated by Blake et al. in 1998[3] and Verloes in 2005.[4] It is a rare syndrome estimated to occur in 1:10,000 to 1:17,000 births, depending on the region.[5,6] CHARGE is an acronym for Coloboma, Heart defects, Atresia of the choanae, Retardation of growth and development, Genital and urinary abnormalities, Ear abnormalities, and/or hearing loss. According to Blake et al.,[3] more precise criteria for diagnosis can be distinguished. They have been divided into three groups: major criteria, minor criteria, and occasional findings. Ear anomalies are included in the major criteria.

According to Blake et al., more than 90% of children with CHARGE syndrome have hearing impairment. The most common defects in the middle ear include abnormalities of the ossicle chain, up to 80%, and aberrant course of the facial nerve, covering almost three-quarters of patients.[7] Inner ear malformations are combinations of cochlea, semicircular canals, and vestibule defects and will be discussed in more detail in this study.

In consequence, the complexity of the previously mentioned defects, together with the frequent co-occurrence of cleft palate (15%–20%) and choanal atresia (50%–60%) in CHARGE patients, may predispose to the poor outcome of conservative treatment.[3] Audiological testing of patients with CHARGE consists of objective tests, including Auditory Brainstem Responses (ABR) and, when available, Auditory Steady-State Responses (ASSR). Audiological diagnosis is carried out early regarding the child's age, allowing for objective audiological tests during physiological, unsedated sleep.

Aim

Our study aimed to describe the spectrum of inner ear malformations in CHARGE syndrome patients and propose a Computed Tomography (CT) detailed scan evaluation methodology. The secondary aim was to correlate the computed tomography findings with hearing thresholds measured with Auditory Steady-State Responses (ASSR).

Methods

Ethical consideration

This retrospective study was approved by the local Institutional Ethics Committee (decision number AKBE/175/2022). The project conforms to The Code of Ethics of the World Medical Association (Declaration of Helsinki).

General study group information

This study performed a retrospective analysis of data obtained from the medical records of patients diagnosed with CHARGE syndrome. Twenty temporal bones of ten consecutive patients diagnosed with the syndrome were subjected to detailed CT analysis focusing on the inner ear and internal acoustic canal. One patient did not have a complete set of audiological tests; however, he was included in the study group due to the CT findings worth detailed radiological assessment. Two independent, experienced investigators (also authors of this manuscript) evaluated the patient's CT scans using the same study protocol and questionnaire.

The patients were diagnosed in two large co-working tertiary centers due to a typical CHARGE combination of clinical features. One center provides genetic and audiological testing and is a full children's tertiary hospital (covering all specialties needed for CHARGE syndrome patients and more). The second one specializes in audiological testing and hearing prostheses, including cochlear implants in children. According to the literature, about 60% of patients with CHARGE present autosomal dominant mutations in the CHD7 gene. More than 1000 mutation variants that can cause CHARGE syndrome have been identified.[6,8–12] In this study, six analyzed patients underwent genetic testing confirming the gene mutations. Four were not analyzed genetically due to parents' no agreement with these tests but presented typical specific and recognizable patterns of CHARGE anomalies.

All ten patients underwent CT scanning of the temporal bone. This study included only the medical records of patients with CHARGE syndrome and CT scans. Patients with CHARGE syndrome diagnosis but no CT were not included. Performing full audiological diagnostics and CT scanning of the temporal bone, which in most cases in small children

requires sedation, is not always a priority due to the other more serious components of the syndrome.

Computed tomography scans analysis procedure in detail

A scan analysis protocol scheme was used for CT evaluation, which had proven satisfactory in previous studies. In addition, a questionnaire with key features was created to assist the investigators with CT evaluation and to standardize their assessment and thus enable the comparison of results. Table 1 presents the mentioned questionare, pointing out the key features of computed tomography analysis. An attempt was made to classify the identified malformations using the newest Sennaroglu classification.[13]

All scans were analyzed using RadiAnt DICOM Viewer 2022.1 64-bit (Medixant, Poznan, Poland) with the MultiPlanar Reconstruction (MPR) option.

Initially, in each case, the researcher set the window level to 500 Hounsfield Units (HU) and the window width to 3000 HU. Then, the protocol developed in earlier studies was used as follows. The MPR tool assessed the semicircular canals in three reference planes. Initially, the axial plane in which the Lateral Semicircular Canal (LSC) should show a typical ''signet ring appearance''. Then, investigators searched for the plane of the Anterior Semicircular Canal (ASC, as described by Pöschl) and the Posterior Semicircular Canal (PSC) with common crus.

The next challenge was to identify and describe the cochlea and vestibule. At first, the longitudinal section of the cochlea was set to assess the basal, middle, and apical turns around the modiolus. Afterward, the adjusted axial plane was set at an angle that allowed for the evaluation of the modiolus and basal turn of the cochlea. Subsequently, a perpendicular plane was defined concerning the modified axial plane, enabling the visualization of the modiolus and all three cochlea turns. However, due to the severity of malformations in analyzed patients, the reference points for determining such planes were disturbed, making the analysis much more difficult.

Internal Acoustic Canal (IAC) diameter was assessed in an axial section of CT by performing measurements at the level of the internal acoustic pore. Firstly, a longitudinal axis line was drawn, and then the IAC diameter was measured as a line drawn perpendicularly to the longitudinal axis of the IAC.

For better visualization and understanding of found malformations in the inner ear in analyzed patients with CHARGE syndrome, examples are shown in Figs. 1, 2 and 3.

Audiological assessment

The assessment of the patient's hearing was carried out in a way suitable for their developmental stage and age. Audiological test results of hearing thresholds (ASSR) were available for analysis in 9 patients (18 ears) and were performed at the age between 3 months to 2.5 years. The results confirmed hearing loss. ASSR results for 500, 1000, 2000, and 4000 Hz were analyzed for the purposes of this study. In ASSR, if the threshold for a particular frequency was absent for the highest intensity level tested (120 dBnHL result as no response), its value was set as 130 dBnHL to facilitate the analyses.

Statistical analysis

Statistical analysis was conducted in the STATISTICA program (TIBCO Software Inc. 2017, version 13.3). The data were tested for normality, parametric, and non-parametric criteria. Detailed statistical analysis was performed using Spearman's correlation test. The level of statistical significance was set at $p = 0.05$.

Results

Twenty temporal bones were analyzed. Two males and eight females were among the investigated children with CHARGE syndrome. As mentioned earlier, one patient did not have a complete set of audiological tests in his medical records (ASSR results were missing). However, he was included in the study group due to the findings worth detailed assessment in CT.

Computed tomography findings

The structure of the cochlea was considered in terms of the basal turn and then jointly in terms of the middle and apical turns. Moreover, the absence or presence of modiolus was assessed. Then, the findings were summarily classified to a specific malformation type according to the newest Senneroglu classification.[13] The basal turn was diagnosed as normal in 90% of cases. However, one patient with cochlear aplasia and rudimentary otocysts was also identified. More variety in structure was found in the middle and apical turns. Besides cochlear aplasia and rudimentary otocyst, the following abnormalities were discovered, which are essential concerning the entire bony labyrinth. Respectively: 2 cases (10%) of dysmorphic (cystic) middle and apical turns, 12 cases (60%) of shortened (less than two turns), and 4 cases (20%) of normal, fully developed cochlea. Modiolus was absent in 20% of cases. In the rest of the cases, the modiolus was considered present. Cochlear anomalies identified in analyzed patients are presented in Table 1, and examples are shown in Fig. 1.

Lateral semicircular canal defects were the most common anomaly in analyzed patients with CHARGE syndrome, affecting 100% of cases. The second most common anomaly concerned the absence of the posterior semicircular canal in all temporal bones except one, where the canal was preserved in its dysmorphic form (5%). The superior semicircular canal was affected by a deep anomaly to a slightly lesser extent. Its absence was observed in 13 temporal bones (65%). In 6, it was hypoplastic (30%), and in 1 case – dysmorphic (5%). However, the vestibule itself was normal in 6 cases (30%), hypoplastic in 12 cases (60%) and absent in one case (5%). Also, the vestibule was dilated in one case, which coexisted with cochlear aplasia. Semicircular canals defects and vestibular anomalies identified in analyzed patients are presented in Table 1, and examples are shown in Fig. 2.

Cochlear Aplasia with a Dilated Vestibule (CADV) and rudimentary otocyst was accompanied by the absence of

Table 1 Results of computed tomography of the inner ear detailed evaluation in patients with CHARGE syndrome.

Patient Nº	Ear	Vestibule	Lateral semicircular canal	Posterior semicircular canal	Superior semicircular canal	IAC diameter (mm)	Basal turn	Middle and apical turns	Modiolus	Cochlear malformation type
1	**R**	Normal	Absent	Absent	Absent	6.08	Normal	Shortened, less than 2 turns	Present	Cochlear hypoplasia type III
1	**L**	Normal	Absent	Absent	Absent	3.76	Normal	Shortened, less than 2 turns	Present	Cochlear hypoplasia type III
2	**R**	Hypoplastic	Absent	Absent	Hypoplastic	2.70	Normal	Cystic, hypoplastic	Absent	Cochlear hypoplasia type II
2	**L**	Hypoplastic	Absent	Absent	Absent	2.27	Normal	Cystic, hypoplastic	Absent	Cochlear hypoplasia type II
3	**R**	Hypoplastic	Absent	Absent	Hypoplastic	3.72	Normal	Shortened, less than 2 turns	Present	Cochlear hypoplasia type III
3	**L**	Hypoplastic	Absent	Absent	Hypoplastic	3.22	Normal	Shortened, less than 2 turns	Present	Cochlear hypoplasia type III
4	**R**	Normal	Absent	Absent	Absent	4.45	Normal	Shortened, less than 2 turns	Present	Cochlear hypoplasia type III
4	**L**	Normal	Absent	Absent	Absent	4.22	Normal	Shortened, less than 2 turns	Present	Cochlear hypoplasia type III
5	**R**	Normal	Absent	Absent	Absent	3.46	Normal	Shortened, less than 2 turns	Present	Cochlear hypoplasia type III
5	**L**	Normal	Absent	Absent	Absent	2.86	Normal	Shortened, less than 2 turns	Present	Cochlear hypoplasia type III
6	**R**	Hypoplastic	Absent	Absent	Hypoplastic	3.14	Normal	Normal	Present	Isolated vestibular labyrinth anomaly
6	**L**	Hypoplastic	Absent	Absent	Absent	3.85	Normal	Normal	Present	Isolated vestibular labyrinth anomaly
7	**R**	Hypoplastic	Absent	Absent	Absent	3.59	Normal	Shortened, less than 2 turns	Present	Cochlear hypoplasia type III
7	**L**	Hypoplastic	Absent	Absent	Absent	3.18	Normal	Shortened, less than 2 turns	Present	Cochlear hypoplasia type III
8	**R**	Hypoplastic	Absent	Absent	Absent	3.22	Normal	Normal	Present	Isolated vestibular labyrinth anomaly
8	**L**	Hypoplastic	Absent	Absent	Absent	3.36	Normal	Shortened, less than 2 turns	Present	Cochlear hypoplasia type III
9	**R**	Hypoplastic	Absent	Absent	Hypoplastic	4.51	Normal	Normal	Present	Isolated vestibular labyrinth anomaly
9	**L**	Hypoplastic	Absent	Absent	Hypoplastic	3.77	Normal	Shortened, less than 2 turns	Present	Cochlear hypoplasia type III
10	**R**	Dilated	Absent	Absent	Dysmorphic	0	Aplasia	Aplasia	Aplasia	Cochlear aplasia with a dilated vestibule (CADV)
10	**L**	Rudimentary otocyst	Absent	Absent	Absent	0	Rudimentary otocyst	Rudimentary otocyst	Rudimentary otocyst	Rudimentary otocyst

IAC, Internal Acoustic Canal.

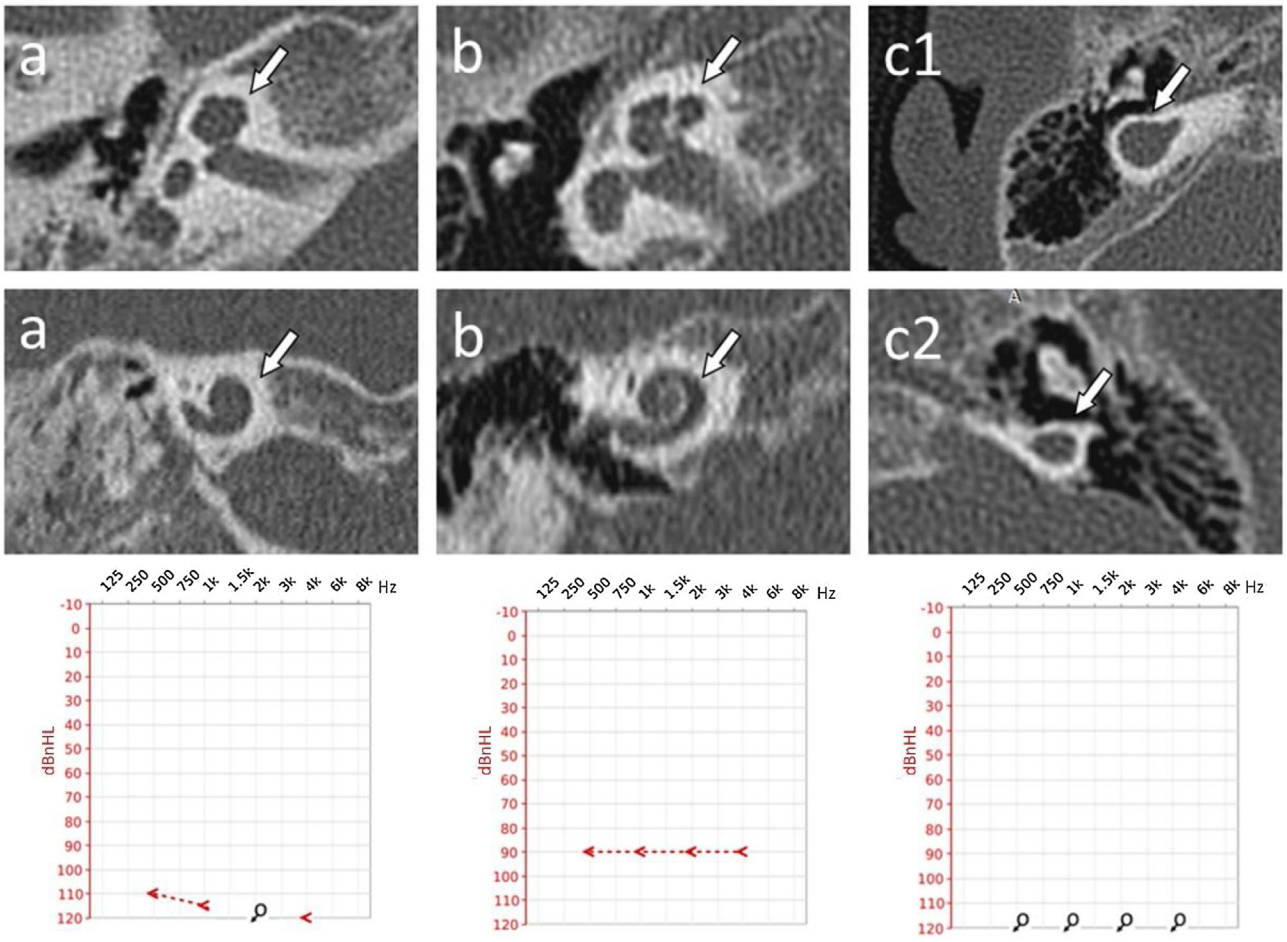

Figure 1 Cochlear malformations identified in computed tomography evaluation and presented in the planes selected for the most precise visualization. Each panel represents a particular patient: (a) Case #2 right ear, cochlear hypoplasia type II; (b) Case #1 right ear, cochlear hypoplasia type III; (c) Case #10 right ear, (c1) Cochlear Aplasia with a Dilated Vestibule (CADV); (c2) Rudimentary otocyst. White arrows indicate anomaly location. Individual cases are supplemented with ASSR reconstruction of audiograms (hearing thresholds in dBnHL), respectively.

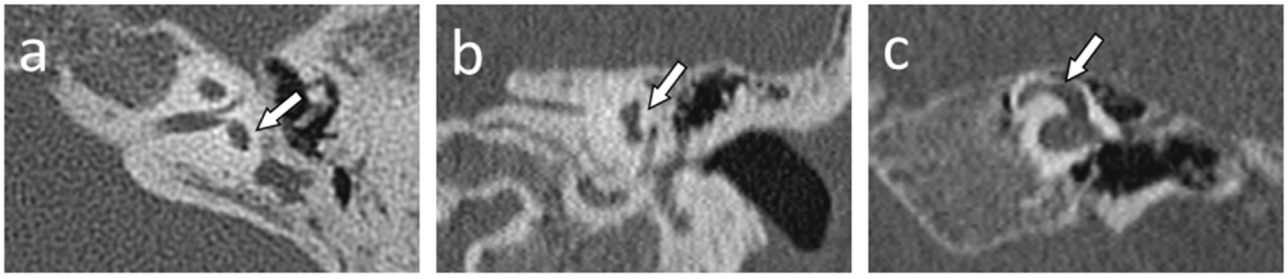

Figure 2 Semicircular canals and vestibule malformations identified in computed tomography evaluation and presented in dedicated planes. Panel a and b) represent the same patient (case #2) with a hypoplastic vestibule and the absence of all three semicircular canals. Panel c (case #10) shows a dysmorphic superior semicircular canal with an accompanying dilated vestibule.

IAC, which for statistical analysis purposes in this study was considered 0.00 mm in diameter. In the remaining cases, the IAC diameter ranged from a minimum dimension of 2.27 mm–6.08 mm. The median IAC diameter of all ears was 3.41 mm, and the Standard Deviation (SD) was 1.37 mm. IAC measurement examples are shown in Fig. 3.

Classification of inner ear malformations

In the final stage of the CT investigation, the analysis of individual elements with a summary aimed at assigning specific malformations to each case according to the Senneroglu classification[13] was carried out. Cochlear Aplasia with a

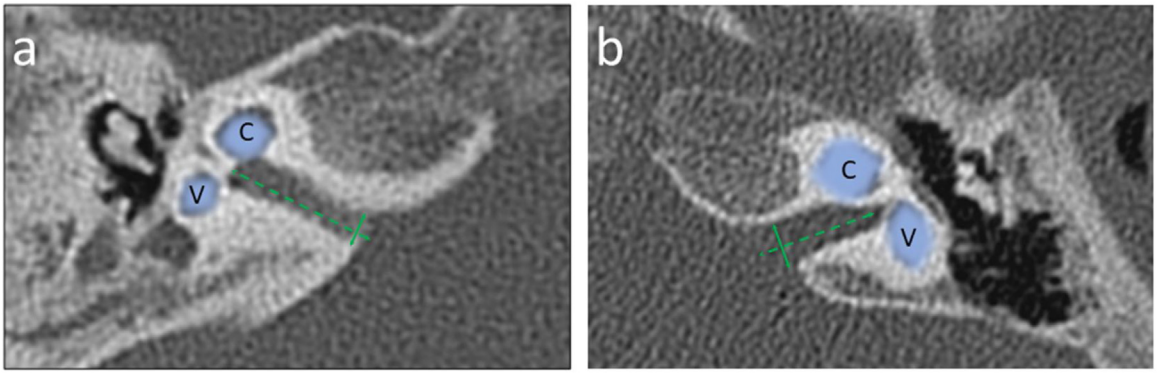

Figure 3 Exemplary measurements of the Internal Auditory Canal (IAC) diameter in an axial section of a computed tomography made at the internal acoustic opening (*porus acusticus internus)*, the IAC diameter (solid line) was determined perpendicular to the longitudinal axis (dashed line) of the IAC; Panel a – narrowed IAC with diameter 2.70 mm (case #2); Panel b – normal sized IAC with diameter 3.76 mm (case #1). Cochlea (C) and Vestibule (V) are marked with a line art over for easier identification.

Dilated Vestibule (CADV) and rudimentary otocyst (Fig. 1c) was seen in both ears in one patient (10% of the analyzed ears, patient #10). It was also related to the almost complete aplasia of the semicircular canals. Only in the right ear was a structure that could be regarded as a remnant of the dysplastic superior semicircular canal (Fig. 2c). Cochlear hypoplasia type II (Fig. 1a) was also seen in both ears in one patient (10% of the analyzed ears, patient #2). Cochlear hypoplasia type III (Fig. 1b) was the most common malformation found in 12 ears (60%). In 4 cases, no cochlear anomaly was identified. Still, in half of them, normal cochlea was accompanied by isolated semicircular canals anomaly and, in two other cases, by a hypoplastic vestibule. In two patients, the defects were not symmetrical in both ears. In both of them, the right ear was more severely affected with noticeable cochlear hypoplasia type III, while the left ear showed only isolated vestibular labyrinth anomaly.

In addition, some correlations were observed between the severity of malformations of individual structures. Firstly, the correlation between the IAC diameter and the severity of vestibular malformations was statistically significant. The more severe the vestibule malformation, the smaller the IAC diameter ($p = 0.013$). In addition, the severity of vestibular defect significantly correlated with the severity of hypoplasia of the basal turn of the cochlea ($p = 0.0057$). Moreover, the correlation occurred in the combination of vestibular malformation with the presence or absence of modiolus. Its absence correlated with the vestibular malformation ($p = 0.0125$).

Audiological findings

Audiological results were unavailable for one patient (patient #6). The remaining nine patients underwent audiological evaluation. The results are shown in Table 2. The ASSR results have been converted to reconstructed audiograms for better understanding and visualization. In the ASSR, there was a statistically significant correlation between the severity of the inner ear malformation and the profoundness of the hearing loss for all of the frequencies, with *p*-values ranging from 0.0001 to 0.0432. The most advanced malformations, such as Cochlear Aplasia with a Dilated Vestibule (CADV) and rudimentary otocyst, showed non-response, which is understandable considering not only the condition of the cochlea (absence) but the absence of IAC on both sides as well. Subsequently, type II hypoplasia correlated with worse hearing level outcomes than type III hypoplasia. Ultimately, patients with the best-developed cochlea had the best audiological results. When inspecting the structure of the cochlea more deeply, taking into consideration the state of the basal, middle, and apical turns and the presence or absence of the modiolus, it was also observed that the better development of these structures, the hearing thresholds become lower (*p*-value ranged from 0.0000 to 0.0432). A significant correlation was also observed for IAC morphology, where absence, narrowing, or normal size correlated statistically significantly with audiological results (*p*-value ranged from 0.0001 to 0.0002). Moreover, a significant correlation with the ASSR scores was observed regarding the exact dimension of the IAC (*p*-value ranged from 0.0076 to 0.0288). The results of the statistical analysis are presented in Table 3.

Discussion

Inner ear malformations are a common finding in individuals diagnosed with CHARGE syndrome. The present study highlights the prevalence and importance of these malformations, shedding light on their diagnostic significance and potential implications for clinical management.

Interestingly, the distribution of malformation types identified in different studies amongst CHARGE syndrome patients is not always convergent. In our study, the dominant malformation is cochlear hypoplasia type III. Less common are cochlear hypoplasia type II, cochlear aplasia, rudimentary otocysts, and isolated defects of the vestibular system. Vesseur et al.[14] analyzed CHARGE syndrome patients and showed more than 60% of cases involved a normal cochlea with isolated semicircular canal defects. In addition, the second significant group was malformations that the researchers described as cochlear hypoplasia type IV, although not without discussion, partly due to a mismatch with any malformations considered. In our study, similar difficulties in identifying the cochlear defect occurred in

Table 2 Auditory Steady-State Response (ASSR) results recorded from analyzed ears – hearing thresholds for frequencies 500, 1000, 2000, and 4000 Hz.

Patient Nº	Cochlear malformation type	Ear	ASSR 500 Hz (dBnHL)	ASSR 1000 Hz (dBnHL)	ASRR 2000 Hz (dBnHL)	ASRR 4000 Hz (dBnHL)
1	Cochlear hypoplasia type III	R	90	90	90	90
1	Cochlear hypoplasia type III	L	100	100	100	100
2	Cochlear hypoplasia type II	R	110	115	NR	120
2	Cochlear hypoplasia type II	L	115	125	NR	NR
3	Cochlear hypoplasia type III	R	65	70	75	75
3	Cochlear hypoplasia type III	L	65	80	75	70
4	Cochlear hypoplasia type III	R	85	100	90	90
4	Cochlear hypoplasia type III	L	35	40	25	15
5	Cochlear hypoplasia type III	R	80	70	60	60
5	Cochlear hypoplasia type III	L	100	100	100	100
6	Isolated vestibular labyrinth anomaly	R	NA	NA	NA	NA
6	Isolated vestibular labyrinth anomaly	L	NA	NA	NA	NA
7	Cochlear hypoplasia type III	R	75	95	70	80
7	Cochlear hypoplasia type III	L	85	90	80	70
8	Isolated vestibular labyrinth anomaly	R	35	50	45	45
8	Cochlear hypoplasia type III	L	75	90	90	90
9	Isolated vestibular labyrinth anomaly	R	45	65	70	70
9	Cochlear hypoplasia type III	L	60	65	70	70
10	Cochlear Aplasia with a Dilated Vestibule (CADV)	R	NR	NR	NR	NR
10	Rudimentary otocyst	L	NR	NR	NR	NR

NR, No Response up to 120 dBnHL; NA, Not Applicable.

case #2. The cochlea was initially classified as incomplete partition type II due to the confluence of the middle and apical turns. Later, with deeper CT evaluation using the proposed protocol in this study, the classification was changed to cochlear hypoplasia type II due to the lack of modiolus and poorly developed basal turn. A similar problem was encountered by Lewis et al., who decided to omit the classic division and classify cochlear hypoplasia into four subgroups: normal, mild, moderate, and severe, with the mild cochlear hypoplasia phenotype as the most common.[11]

In their study group, Aragón-Ramos et al.[15] found cochlear hypoplasia type III as the most popular defect, consistent with our results. Da Costa Monsanto et al.[16] found cochlear hypoplasia in all examined temporal bones from donors with CHARGE syndrome. Before Sennaroglu systematized inner ear malformations in 2017, researchers did not always use a unified description when analyzing defects in CHARGE patients. Rah et al.[17] identified inner ear malformations in 82% of CHARGE patients undergoing cochlear implantation, collectively termed cochlear hypoplasia, which was consistent with similar work of Birman et al.,[18] where broadly interpreted cochlear hypoplasia has been reported in 80% of implanted patients. However, significantly different results are presented by Vincenti et al.[19] In their study, not one of the eight analyzed patients with CHARGE was found to have cochlear hypoplasia. The anomalies included incomplete partition types I and II, common cavity, and, in half of the cases, an isolated defect of the vestibular labyrinth. As their work focused on aspects of cochlear implantation, they did not present the methodology of CT evaluation used for assessing the inner ear structures.

The genetic background also seems to be not without significance in the following considerations. According to Zentner et al.,[9] inner ear anomalies identified through temporal bone CT or skull X-Ray were significantly higher in individuals with CHD7 mutations (98%) than those without mutations (75%). Adding a genetic factor to the description of malformations could shed light on the observed discrepancies.

In all patients in our study group, the absence of semicircular canals was found to some extent. It aligns with other authors who even argued that it is a reliable and crucial factor for diagnostic purposes. According to Lalani et al.,[20] a combination of coloboma, choanal atresia, and abnormal semicircular canals highly predicted the presence of a CHD7 mutation. Wineland et al.[21] found semicircular canal hypoplasia/aplasia more prevalent than other characteristic features such as coloboma or choanal atresia in their study population. Sanlaville et al.[22] showed that the absence of a semicircular canal is detectable by ultrasound and should be searched for in fetuses with conotruncal heart defects. Moreover, in 10 fetuses with CHD7 mutations, arhinencephaly and semicircular canal agenesis were

Table 3 Spearman rank correlation results between inner ear anomaly severity and hearing thresholds determined in Auditory Steady-State potentials (ASSR) for 500, 1000, 2000, and 4000 Hz.

Analyzed pair of variables	Number of ears	R Spearman	*p*-value
IAC (Absent-1/Narrow-2/Normal-3) & ASSR 500 Hz	18	−0.7788	**0.0001***
IAC (Absent-1/Narrow-2/Normal-3) & ASSR 1000 Hz	18	−0.7703	**0.0002***
IAC (Absent-1/Narrow-2/Normal-3) & ASRR 2000 Hz	18	−0.7713	**0.0002***
IAC (Absent-1/Narrow-2/Normal-3) & ASRR 4000 Hz	18	−0.7775	**0.0001***
IAC diameter (mm) & ASSR 500 Hz	18	−0.5927	**0.0095***
IAC diameter (mm) & ASSR 1000 Hz	18	−0.6068	**0.0076***
IAC diameter (mm) & ASRR 2000 Hz	18	−0.5705	**0.0134***
IAC diameter (mm) & ASRR 4000 Hz	18	−0.5149	**0.0288***
Basal turn (Aplasia-1/R-otocyst -2/Normal-3) & ASSR 500 Hz	18	−0.5459	**0.0191***
Basal turn (Aplasia-1/R-otocyst -2/Normal-3) & ASSR 1000 Hz	18	−0.5473	**0.0187***
Basal turn (Aplasia-1/R-otocyst -2/Normal-3) & ASRR 2000 Hz	18	−0.4812	**0.0432***
Basal turn (Aplasia-1/R-otocyst -2/Normal-3) & ASRR 4000 Hz	18	−0.5153	**0.0286***
Middle and apical turns (Aplasia-1/R-otocyst-2/Dysmo-3/Shortened-4/Normal-5) & ASSR 500 Hz	18	−0.8138	**0.0000***
Middle and apical turns (Aplasia-1/R-otocyst-2/Dysmo-3/Shortened-4/Normal-5) & ASSR 1000 Hz	18	−0.7986	**0.0001***
Middle and apical turns (Aplasia-1/R-otocyst-2/Dysmo-3/Shortened-4/Normal-5) & ASRR 2000 Hz	18	−0.7663	**0.0002***
Middle and apical turns (Aplasia-1/R-otocyst-2/Dysmo-3/Shortened-4/Normal-5) & ASRR 4000 Hz	18	−0.7621	**0.0002***
Modiolus (Absent-1/Present-2) & ASSR 500 Hz	18	−0.7234	**0.0007***
Modiolus (Absent-1/Present-2) & ASSR 1000 Hz	18	−0.7253	**0.0007***
Modiolus (Absent-1/Present-2) & ASRR 2000 Hz	18	−0.7288	**0.0006***
Modiolus (Absent-1/Present-2) & ASRR 4000 Hz	18	−0.7284	**0.0006***
Cochlear malformation type (Aplasia-1/R-otocyst2/Hypopl-II-3/Hypopl-III-4/Normal-5) & ASSR 500 Hz	18	−0.8138	**0.0000***
Cochlear malformation type (Aplasia-1/R-otocyst2/Hypopl-II-3/Hypopl-III-4/Normal-5) & ASSR 1000 Hz	18	−0.7986	**0.0001***
Cochlear malformation type (Aplasia-1/R-otocyst2/Hypopl-II-3/Hypopl-III-4/Normal-5) & ASRR 2000 Hz	18	−0.7663	**0.0002***
Cochlear malformation type (Aplasia-1/R-otocyst2/Hypopl-II-3/Hypopl-III-4/Normal-5) & ASRR 4000 Hz	18	−0.7621	**0.0002***

The ''*p*'' value represents the level of significance. The asterisks (*) and bold font were used to mark statistically significant correlations ($p < 0.05$).
R-otocyst, Rudimentary Otocyst; Dysmo, Dysmorphic; Hypopl-II, Hypoplasia type II; Hypopl-III, Hypoplasia type III.

two constant features. Kimura and Kaga[23] found that children with CHARGE and semicircular canal aplasia presented severe dysfunction of the vestibular ocular reflex, which pointed to impaired functioning of the vestibular organ, which in turn may be associated with impaired motor development.

According to other studies in the literature, the width of the IAC correlates with the occurrence of inner ear malformations and bilateral sensorineural hearing loss.[24,25] Another study[16] revealed that the width of the IAC in individuals with inner ear malformations did not show a significant correlation with the number of Spiral Ganglion Neurons (SGNs) or the presence of aplastic/hypoplastic cochlear nerves. Our study considered a narrowed IAC less than 3 mm wide. However, in some studies in the literature,[24–26] sometimes the IAC width of 2 mm is considered the lower limitation of normal size.

All of the above mentioned studies from the literature, except for one, were based on small groups of patients due to the low prevalence of the discussed syndrome. The number of patients ranged from 6 to 13 cases. Only the Vesseur et al. study[14] included a larger group of 42 patients. Still, the data analysis also included analog CT (data dating back to 1996), in which multiplanar reconstruction was impos-

sible. The sample size was also a limitation of our study. A series of 20 ears could influence the generalizability of the findings, making it necessary to conduct larger, preferably multi-center studies to validate the prevalence of inner ear malformations in a broader population of individuals diagnosed with CHARGE syndrome. Further investigations exploring the specific molecular mechanisms underlying these malformations could provide valuable insights into the developmental processes disrupted in CHARGE syndrome.

Conclusions

The results of this study demonstrate that rudimentary semicircular canals or a complete absence of these canals were universally observed in all patients diagnosed with CHARGE syndrome. This finding supports the idea that inner ear anomalies are a hallmark feature of the CHARGE syndrome, contributing to its distinct clinical profile.

The presence of inner ear malformations has substantial clinical implications. Audiological assessments are crucial for individuals with CHARGE syndrome, as hearing loss is a common feature associated with inner ear anomalies. Early detection of these malformations can guide appropriate interventions, such as hearing aids or cochlear implants, which may significantly improve developmental outcomes and communication for affected individuals. Recognizing inner ear malformations as a diagnostic criterion presents implications beyond clinical diagnosis. A better understanding of these malformations can advance the knowledge of CHARGE syndrome pathophysiology. It may also help guide future research into targeted therapies to mitigate the impact of inner ear anomalies on hearing and balance function. This study adds to the knowledge and diagnostics of patients with CHARGE syndrome for healthcare use.

Declaration of Generative AI and AI-assisted technologies in the writing process

While preparing this work, the authors did not use AI or AI-assisted technologies.

Conflicts of interest

The authors declare no conflict of interest, including any financial interest or support related to this manuscript. This research did not receive any specific grant from the public, commercial, or not-for-profit funding agencies.

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Wnioski

W publikacjach stanowiących cykl rozprawy doktorskiej zaproponowano i szczegółowo opisano protokół metodologii rekonstrukcji i analizy obrazów tomografii komputerowej kości skroniowych oraz jej zastosowanie do oceny wad wrodzonych ucha wewnętrznego w praktyce klinicznej. Protokół powstał w oparciu o wielopłaszczyznową rekonstrukcję tomografii komputerowej (CT). Pierwszym krokiem był wybór poziomu okna na 500 jednostek Hounsfielda (HU) i szerokości na 3000 HU. Ustalono trzy płaszczyzny dedykowane trzem kanałom półkolistym oraz analogiczne trzy płaszczyzny dotyczące ślimaka. Płaszczyzny, w których oceniano ślimak, zostały opracowane mając na celu uwidocznienie jego struktury zewnętrzenej - morfologii zakrętu podstawnego, środkowego i szczytowego, oraz architektury wewnętrznej – morfologii wrzecionka. Tak dobrane płaszczyzny rekonstrukcji umożliwiły powtarzalny schemat oceny. Aby w pełni zrozumieć różnorodność wad rozwojowych ucha wewnętrznego, wykonano i poddano wnikliwej analizie rekonstrukcje 3D. W przedstawianym cyklu publikacji przedstawiono także wartość zastosowania proponowanego rozwiązania analizy obrazów CT uszu w praktyce klinicznej.

W przypadku implantacji ślimakowej, zdefiniowanie budowy ślimaka przed planowaną operacją ułatwia chirurgowi wybór rodzaju elektrody oraz szczegółowe zaplanowanie operacji. Ponadto, jak wykazano, pozwala z dużym prawdopodobieństwem przewidzieć śródoperacyjne zdarzenia nieporządane takie jak niekontrolowany wypływ płynu mózgowo-rdzeniowego (*gusher*) lub niepełna insercja elektrody. Dodatkowo wykazano, iż pacjenci z malformacjami ucha wewnętrznego wymagają bardziej indywidualnego podejścia do operacji i rehabilitacji oraz wyższych intensywności stymulacji, aby uzyskać wrażenia dźwiękowe. Zastosowanie opracowanego protokołu rekonstrukcji i analizy obrazów CT ucha wewnętrznego pozwala na zwiększenie bezpieczeństwa przeprowadzenia wszczepienia implantu ślimakowego u pacjentów z wrodzonymi wadami ucha wewnętrznego.

Kolejnym zagadnieniem podejmowanym w przedstawianym cyklu rozprawy doktorskiej są anomalie rozwojowe dotyczące ucha wewnętrznego u pacjentów z zespołem CHARGE. W trzeciej publikacji z cyklu, wykorzystując opracowany wcześniej protokół oceny CT, zidentyfikowano liczne nieprawidłowości rozwojowe w obrębie kości skroniowych pacjentów z tym zespołem. Stwierdzono, iż stopień zaawansowania wady ślimaka, jak również stopień rozwoju przewodu słuchowego wewnętrznego korelował z wynikami badań

audiologicznych, tj. im bardziej zaburzona architektura ślimaka lub im mniejsza średnica IAC tym bardziej podwyższone progi słyszenia.

Analiza dotycząca narządu przedsionkowego u pacjentów z zespołem CHARGE ujawniła stały element malformacji rozwojowej jakim był brak kanału półkolistego bocznego, z którym często współistniały malformacje kanałów półkolistych tylnego i górnego. Istotne statystycznie korelacje występowały między nasileniem deformacji przedsionka a średnicą IAC, nasileniem hipoplazji zakrętu podstawnego ślimaka oraz obecnością lub brakiem wrzecionka. Tym samym dowiedziono, iż malformacja dotycząca jednego z elementów architektury kości skroniowej, w znaczącej większości pociąga za sobą malformacje pozostałych struktur. Stanowi to spójny obraz wad ucha wewnętrznego będących jednym z dużych kryteriów rozpoznania zespołu CHARGE. Opracowane w publikacji wyniki badań audiologicznych, korelujące ze stopniem nasilenia malformacji ślimaka, są kluczowe do podejmowania decyzji dotyczących dalszej rehabilitacji narządu słuchu. Wybór metody protezowania słuchu u pacjentów z wadami ucha wewnętrznego zależy bezpośrednio od architektury ślimaka i wyników badań audiologicznych, a pośrednio od stanu ogólnego pacjenta i współtowarzyszących obciążeń dotyczacych innych układów.

Przedstawione publikacje stanowiące cykl rozprawy doktorskiej wnoszą istotny wkład w dziedzinie audiologii i otologii do diagnostyki i leczenia niedosłuchu poprzez nowatorskie opracowanie i szczegółowe przedstawienie protokołu metodologii rekonstrukcji i analizy obrazów tomografii komputerowej oraz jego zastosowania do oceny wad wrodzonych ucha wewnętrznego w praktyce klinicznej. Przedstawiona jest również korelacja szczegółów anatomicznych malformacji ucha wewnętrznego z wynikami audiologicznymi. Co za tym idzie, wiedza uzyskana z tak opracowanych obrazów TK kości skroniowych jest istotna z praktycznego punktu widzenia i może być bardzo przydatna do dalszych badań i opracowywania bardziej precyzyjnych i zindywidualizowanych metod leczenia niedosłuchu w grupie pacjentów z wadami wrodzonymi ucha wewnętrznego.

Piśmiennictwo

Spis piśmiennictwa dostępny na końcu każdej z trzech publikacji prezentowanego cyklu rozprawy doktorskiej.

Oświadczenia wszystkich współautorów cyklu publikacji stanowiących rozprawę doktorską

Warszawa, 29.08.2024
.....
(miejscowość, data)

Prof. dr hab.n.med. Magdalena Lachowska
.....
(imię i nazwisko)

OŚWIADCZENIE

Jako współautor pracy pt. „Anatomical and clinical aspects and outcomes of bilateral cochlear implantation in cochlear hypoplasia type IV – a case report" oświadczam, iż mój własny wkład merytoryczny w przygotowanie, przeprowadzenie i opracowanie badań oraz przedstawienie pracy w formie publikacji stanowi: przygotowanie projektu badań, zebranie i opracowanie danych, przygotowanie manuskryptu, opracowanie literatury, pełnienie funkcji autora korespondencyjnego artykułu.

Mój udział procentowy w przygotowaniu publikacji określam jako 15 %.

Wkład lek. Agaty Szleper w powstawanie publikacji określam jako 70 %,
obejmował on: przygotowanie projektu badań i metodologii, zebranie i opracowanie danych, przygotowanie manuskryptu, opracowanie literatury, nadzorowanie prac zespołu.

Jednocześnie wyrażam zgodę na wykorzystanie w/w pracy jako część rozprawy doktorskiej lek. Agaty Szleper

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(podpis oświadczającego)

Warszawa, 29.08.2024

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(miejscowość, data)

Prof. dr hab.n.med. Magdalena Lachowska

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(imię i nazwisko)

OŚWIADCZENIE

Jako współautor pracy pt. „Computed tomography multi-planar and 3D image assessment protocol for detailed analysis of inner ear malformations in patients undergoing cochlear implantation counseling" oświadczam, iż mój własny wkład merytoryczny w przygotowanie, przeprowadzenie i opracowanie badań oraz przedstawienie pracy w formie publikacji stanowi: przygotowanie projektu badań, zebranie i interpretacja danych, przygotowanie manuskryptu, opracowanie literatury, pełnienie funkcji autora korespondencyjnego artykułu.

Mój udział procentowy w przygotowaniu publikacji określam jako 12 %.

Wkład lek. Agaty Szleper w powstawanie publikacji określam jako 75 %,
obejmował on: przygotowanie projektu badań i metodologii, zebranie i interpretacja danych, przygotowanie manuskryptu, opracowanie literatury, nadzorowanie prac zespołu.

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Warszawa, 29.08.2024

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(imię i nazwisko)

OŚWIADCZENIE

Jako współautor pracy pt. „Detailed analysis of inner ear malformations in CHARGE syndrome patients - correlation with audiological results and proposal for computed tomography scans evaluation methodology" oświadczam, iż mój własny wkład merytoryczny w przygotowanie, przeprowadzenie i opracowanie badań oraz przedstawienie pracy w formie publikacji stanowi: nadzór merytoryczny nad treścią artykułu, nadzór nad przeprowadzeniem badań, nadzór nad interpretacją otrzymanych wyników badań, analiza statystyczna, pisanie artykułu, pełnienie funkcji autora korespondencyjnego artykułu.

Mój udział procentowy w przygotowaniu publikacji określam jako 10 %.

Wkład lek. Agaty Szleper w powstawanie publikacji określam jako 80 %,
obejmował on: opracowanie metodologii, zabranie materiałów badawczych i bibliografii, opracowanie materiału do analiz statystycznych, szczegółową analizę i interpretację wyników, wizualizację wyników, pisanie artykułu, nadzorowanie prac zespołu.

Jednocześnie wyrażam zgodę na wykorzystanie w/w pracy jako część rozprawy doktorskiej lek. Agaty Szleper.

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(podpis oświadczającego)

WARSZAWA, 8.08.2024
(miejscowość, data)

TOMASZ WOJCIECHOWSKI
(imię i nazwisko)

OŚWIADCZENIE

Jako współautor pracy pt. „Anatomical and clinical aspects and outcomes of bilateral cochlear implantation in cochlear hypoplasia type IV – a case report" oświadczam, iż mój własny wkład merytoryczny w przygotowanie, przeprowadzenie i opracowanie badań oraz przedstawienie pracy w formie publikacji stanowi: nadzór merytoryczny nad treścią artykułu, interpretacja otrzymanych wyników badań, wizualizacja wyników badań.

Mój udział procentowy w przygotowaniu publikacji określam jako 4 %.

Wkład lek. Agaty Szleper w powstawanie publikacji określam jako 70 %,
obejmował on: opracowanie metodologii, zebranie materiałów badawczych i bibliografii, szczegółową analizę i interpretację wyników, pisanie artykułu, nadzorowanie prac zespołu.

Jednocześnie wyrażam zgodę na wykorzystanie w/w pracy jako część rozprawy doktorskiej lek. Agaty Szleper

Tomasz Wojciechowski
(podpis oświadczającego)

Warszawa, 8.08.2024
(miejscowość, data)

TOMASZ WOJCIECHOWSKI
(imię i nazwisko)

OŚWIADCZENIE

Jako współautor pracy pt. „Detailed analysis of inner ear malformations in CHARGE syndrome patients - correlation with audiological results and proposal for computed tomography scans evaluation methodology" oświadczam, iż mój własny wkład merytoryczny w przygotowanie, przeprowadzenie i opracowanie badań oraz przedstawienie pracy w formie publikacji stanowi: opracowanie metodologii, interpretacja wyników badań, nadzór merytoryczny nad treścią artykułu, nadzór nad interpretacją otrzymanych wyników badań.

Mój udział procentowy w przygotowaniu publikacji określam jako 9 %.

Wkład lek. Agaty Szleper w powstawanie publikacji określam jako 80 %,
obejmował on: opracowanie metodologii, zabranie materiałów badawczych i bibliografii, opracowanie materiału do analiz statystycznych, szczegółową analizę i interpretację wyników, wizualizację wyników, pisanie artykułu, nadzorowanie prac zespołu.

Jednocześnie wyrażam zgodę na wykorzystanie w/w pracy jako część rozprawy doktorskiej lek. Agaty Szleper.

Tomasz Wojciechowski
(podpis oświadczającego)

WARSZAWA, 8.08.2024
(miejscowość, data)

TOMASZ WOJCIECHOWSKI
(imię i nazwisko)

OŚWIADCZENIE

Jako współautor pracy pt. „Computed tomography multi-planar and 3D image assessment protocol for detailed analysis of inner ear malformations in patients undergoing cochlear implantation counseling" oświadczam, iż mój własny wkład merytoryczny w przygotowanie, przeprowadzenie i opracowanie badań oraz przedstawienie pracy w formie publikacji stanowi: opracowanie metodologii, nadzór merytoryczny nad treścią artykułu, analiza i interpretacja otrzymanych wyników badań, wizualizacja wyników badań.

Mój udział procentowy w przygotowaniu publikacji określam jako 12 %.

Wkład lek. Agaty Szleper w powstawanie publikacji określam jako 75 %,
obejmował on: opracowanie metodologii oraz zebranie bibliografii, szczegółową analizę, interpretację i przedstawienie wyników badań, pisanie artykułu, nadzorowanie prac zespołu.

Jednocześnie wyrażam zgodę na wykorzystanie w/w pracy jako część rozprawy doktorskiej lek. Agaty Szleper.

Tomasz Wojciechowski
(podpis oświadczającego)

Poznań, 09.09.2024

miejscowość, data

DR. HAB. N. MED.
KATARZYNA HARMCKA-PROMCKA

imię i nazwisko

OŚWIADCZENIE

Jako współautor pracy pt. „Detailed analysis of inner ear malformations in CHARGE syndrome patients - correlation with audiological results and proposal for [illegible] tomography scans evaluation methodology" oświadczam, iż mój własny wkład merytoryczny w opracowanie, przeprowadzenie i opracowanie badań oraz przedstawienie pracy w formie publikacji stanowi: udostępnienie materiału poddanego analizie w artykule.

Mój udział procentowy w przygotowaniu publikacji określam jako 1 %.

Wkład lek. Agaty Szleper w powstawanie publikacji określam jako 80 %,
obejmował on: opracowanie metodologii, zabranie materiałów badawczych i bibliografii, opracowanie materiału do analiz statystycznych, szczegółową analizę i interpretację wyników, wizualizację wyników, pisanie artykułu, nadzorowanie prac zespołu.

Jednocześnie wyrażam zgodę na wykorzystanie w/w pracy jako część rozprawy doktorskiej lek. Agaty Szleper

podpis oświadczającego

26.08.2024
(miejscowość, data)

AGNIESZKA PASTUSZKA
(imię i nazwisko)

OŚWIADCZENIE

Jako współautor pracy pt. „Anatomical and clinical aspects and outcomes of bilateral cochlear implantation in cochlear hypoplasia type IV – a case report" oświadczam, iż mój własny wkład merytoryczny w przygotowanie, przeprowadzenie i opracowanie badań oraz przedstawienie pracy w formie publikacji stanowi: nadzór nad interpretacją wyników badań, pisanie artykułu.

Mój udział procentowy w przygotowaniu publikacji określam jako 5 %.

Wkład lek. Agaty Szleper w powstawanie publikacji określam jako 70 %,
obejmował on: opracowanie metodologii, zebranie materiałów badawczych i bibliografii, szczegółową analizę i interpretację wyników, pisanie artykułu, nadzorowanie prac zespołu.

Jednocześnie wyrażam zgodę na wykorzystanie w/w pracy jako część rozprawy doktorskiej lek. Agaty Szleper.

A. Pastuszka
(podpis oświadczającego)

Warszawa, 13.09.2024
(miejscowość, data)

Zuzanna Tuliszewska-Kłoczyńska
(imię i nazwisko)

OŚWIADCZENIE

Jako współautor pracy pt. „Anatomical and clinical aspects and outcomes of bilateral cochlear implantation in cochlear hypoplasia type IV – a case report" oświadczam, iż mój własny wkład merytoryczny w przygotowanie, przeprowadzenie i opracowanie badań oraz przedstawienie pracy w formie publikacji stanowi: nadzór nad interpretacją wyników badań, pisanie artykułu.

Mój udział procentowy w przygotowaniu publikacji określam jako 5 %.

Wkład lek. Agaty Szleper w powstawanie publikacji określam jako 70 %,
obejmował on: opracowanie metodologii, zebranie materiałów badawczych i bibliografii, szczegółową analizę i interpretację wyników, pisanie artykułu, nadzorowanie prac zespołu.

Jednocześnie wyrażam zgodę na wykorzystanie w/w pracy jako część rozprawy doktorskiej lek. Agaty Szleper.

(podpis oświadczającego)

Warszawa, 29.08.2024r.
(miejscowość, data)

prof. dr hab. n. med. Kazimierz Niemczyk
(imię i nazwisko)

OŚWIADCZENIE

Jako współautor pracy pt. „Anatomical and clinical aspects and outcomes of bilateral cochlear implantation in cochlear hypoplasia type IV – a case report" oświadczam, iż mój własny wkład merytoryczny w przygotowanie, przeprowadzenie i opracowanie badań oraz przedstawienie pracy w formie publikacji stanowi: udostępnienie miejsca i sprzętu do przeprowadzenia badań naukowych, nadzór merytoryczny nad przeprowadzanym badaniami.

Mój udział procentowy w przygotowaniu publikacji określam jako 1 %.

Wkład lek. Agaty Szleper w powstawanie publikacji określam jako 70 %,
obejmował on: opracowanie metodologii, zebranie materiałów badawczych i bibliografii, szczegółową analizę i interpretację wyników, pisanie artykułu, nadzorowanie prac zespołu.

Jednocześnie wyrażam zgodę na wykorzystanie w/w pracy jako część rozprawy doktorskiej lek. Agaty Szleper.

KIEROWNIK
Katedry i Kliniki Otorynolaryngologii,
Chirurgii Głowy i Szyi
prof. dr hab. n. med. Kazimierz Niemczyk

(podpis oświadczającego)

Warszawa 29.08.2024 r.
(miejscowość, data)

prof. dr hab. n. med. Kazimierz Niemczyk
(imię i nazwisko)

OŚWIADCZENIE

Jako współautor pracy pt. „Computed tomography multi-planar and 3D image assessment protocol for detailed analysis of inner ear malformations in patients undergoing cochlear implantation counseling" oświadczam, iż mój własny wkład merytoryczny w przygotowanie, przeprowadzenie i opracowanie badań oraz przedstawienie pracy w formie publikacji stanowi: udostępnienie miejsca i sprzętu do przeprowadzenia badań naukowych, nadzór merytoryczny nad przeprowadzanym badaniami.

Mój udział procentowy w przygotowaniu publikacji określam jako 1 %.

Wkład lek. Agaty Szleper w powstawanie publikacji określam jako 75 %,
obejmował on: opracowanie metodologii oraz zebranie bibliografii, szczegółową analizę, interpretację i przedstawienie wyników badań, pisanie artykułu, nadzorowanie prac zespołu.

Jednocześnie wyrażam zgodę na wykorzystanie w/w pracy jako część rozprawy doktorskiej lek. Agaty Szleper.

KIEROWNIK
Katedry i Kliniki Otorynolaryngologii,
Chirurgii Głowy i Szyi

.....prof. dr hab. n. med. Kazimierz Niemczyk
(podpis oświadczającego)