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先天性巨大中耳胆脂瘤 合并中耳畸形个案研究

Congenital Giant Middle Ear Cholesteatoma Combined with Middle Ear Malformation: A Case Report

唐语祺¹ 何伟平² 郭华民² 刘景²

TANG Yu-qi, HE Wei-ping, GUO Hua-min, LIU Jing

【摘要】目的 先天性巨大中耳胆脂瘤合并中耳畸形由于病变位置隐匿、早期症状不典型,易因误诊而延误最佳治疗时机。本文通过分析病例临床资料,系统总结该病的临床特点,回顾耳显微镜下乳突改良根治手术的要点,综合国内外文献探讨该疾病的发病机制,旨在提高临床医师对该病诊断、鉴别诊断的认知能力及个体化手术策略制订,强调影像学在术前评估中的价值与选择合适手术方式的重要意义。**方法** 报道1例7岁男性患儿因右耳反复流脓2年就诊,曾多次诊断为急性中耳炎,抗感染治疗效果不佳。于广州中医药大学第一附属医院耳鼻喉科完善检查后明确诊断为先天性巨大中耳胆脂瘤合并中耳畸形。术前评估为Potsic IV期。经术前讨论及家属知情同意,患儿接受耳显微镜下乳突改良根治术,在保护原有解剖结构的情况下彻底清除病变组织。**结果** 术后病理证实胆脂瘤诊断,随访患儿无耳鸣、眩晕、面瘫等术后并发症,可正常交流,暂无2次手术计划。**结论** 首要表现为耳道流脓而无面痛、面瘫、眩晕的先天性中耳胆脂瘤易误诊为中耳炎,颞骨高分辨率计算机断层扫描、中耳磁共振成像与弥散加权成像等影像学对该病的早期定位、范围评估和治疗方案选择具有指导意义。在明确先天性胆脂瘤诊断后应尽快采取合适的手术方式彻底清除病变,尽可能保留原有结构,减少病变对周围组织及相关功能的不可逆损害,最大程度提高患者预后及远期生活质量。

【关键词】 先天性中耳胆脂瘤;先天性中耳畸形;乳突改良根治术**【中图分类号】** R764.2**【文献标识码】** A**【文章编号】** 1672-4933(2026)05-0501-06

【Abstract】 Objective This study aims to enhance the diagnostic capability of clinicians in diagnosis and differential diagnosis, promote individualized surgical strategy development, and emphasize the value of imaging in preoperative evaluation and the importance of selecting appropriate surgical approaches, by analyzing the clinical data of a case of congenital giant middle ear cholesteatoma with middle ear malformation, which is prone to misdiagnosis due to its concealed location and atypical early symptoms, often resulting in delayed optimal treatment; this study systematically summarizes the clinical features, reviews the key aspects of modified radical mastoidectomy under otomicroscopy, and discusses the pathogenesis with reference to domestic and international literature. **Methods** This report describes a 7-year-old male who presented with recurrent right otorrhea for 2 years and had received repeated diagnoses of acute otitis media with a poor response to anti-infective therapy; however, following comprehensive examination at our institution, the patient was definitively diagnosed with congenital giant middle ear cholesteatoma with middle ear malformation, and preoperative staging was determined as Potsic stage IV. After thorough preoperative discussion and with informed consent obtained from the family, the patient underwent modified radical mastoidectomy under otomicroscopy, achieving complete lesion removal while preserving the original anatomical structures. **Results** Postoperative histopathological examination confirmed the diagnosis of cholesteatoma. During follow-up, the patient had no postoperative complications such as tinnitus, vertigo, or facial palsy, and demonstrated normal communication ability; no second-stage surgical intervention is currently planned. **Conclusion** Congenital middle ear cholesteatoma with otorrhea as the primary manifestation, but without facial pain, facial palsy, or vertigo, is susceptible to misdiagnosis as otitis media. Imaging modalities including high-resolution CT of the temporal bone, middle ear MRI, and diffusion-weighted imaging are indispensable for early localization, extent assessment, and treatment planning. Once the diagnosis has been established, appropriate surgical intervention should be performed promptly to achieve complete removal of the lesion, preserve native structures to the greatest extent, minimize irreversible damage to surrounding tissues and functions, and thus improve the long-term prognosis and quality of life.

【Key words】 Congenital middle ear cholesteatoma; Congenital middle ear malformation; Modified radical mastoidectomy

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作者单位:1 广州中医药大学第一临床医学院 广州 510499

2 广州中医药大学第一附属医院耳鼻喉科 广州 510405

作者简介:唐语祺 硕士 住院医师;研究方向:耳科学

通讯作者:刘景, E-mail: weather8lj@126.com

中耳胆脂瘤(middle ear cholesteatoma)由基质上皮及其周围组织、角蛋白碎片组成,其异常生长会侵蚀破坏周围组织,累及听觉及前庭器官,甚至危及生命^[1]。先天性胆脂瘤(congenital cholesteatoma, CC)起源于角化上皮,因胚胎发育过程中外胚层上皮组织残留于颞骨逐渐堆积生长形成。目前诊断CC需符合由Derlacki等^[2]1965年提出,经Levenson等^[3]修订的标准,主要包括:①鼓膜完整,鼓膜内侧可见白色珍珠样新生物;②无耳流脓病史;③无耳部手术史。本文报道1例先天性巨大胆脂瘤合并中耳畸形患儿,因其年龄、病史的特殊性及其病情的反复性,曾被反复漏诊。笔者团队通过丰富的临床经验、精准的术前诊断、合理的手术方式,彻底清除病变,保护了重要结构及功能,术后患儿无面瘫、眩晕等并发症,恢复良好。本研究通过广州中医药大学第一附属医院医学伦理委员会审批(K-2026-206),并获得患者监护人知情同意。

1 资料与方法

1.1 临床资料

患儿男性,7岁,因右耳反复流脓2年至广州中医药大学第一附属医院耳鼻喉科就诊。近2年内右耳流脓、疼痛,曾多次于外院就诊,诊断为慢性中耳炎,口服抗生素及药物滴耳后暂时好转,未接受其他如鼓膜置管等干预,未行听力测试,症状反复发作。既往听力初筛通过,无外伤及耳部手术史。辅助检查:纯音听阈示右耳平均听阈(PTA)52 dB HL,气骨导差(air-bone gap, ABG)35 dB HL,左耳PTA 18 dB HL(图1)。耳内镜提示右外耳道狭窄,后壁可疑骨性凸起,深部见分泌物,下方鼓膜无法窥及(图2)。声导抗检查示右耳鼓室图未引出,镫骨肌反射未引出。左耳鼓室图A型(图3)。视频头脉冲试验提示

各半规管高频功能增益正常(图4)。面神经电图提示右侧面神经无损伤。颞骨高分辨率计算机断层扫描(high resolution computed tomography, HRCT)显示右鼓室及乳突窦骨质部分破坏,内有大量软组织影填充,累及骨性外耳道后壁,外耳道局部狭窄。部分听骨链、后半规管、外半规管破坏,鼓室天盖缺损(图5)。中耳增强磁共振成像(magnetic resonance imaging, MRI)见乳突窦扩大,胆脂瘤自乳突延伸至鼓室,软组织填充大小约33 mm×17 mm×18 mm,天盖破坏,硬脑膜暴露,耳蜗及内听道口结构正常。病变部位增强扫描强化不明显,弥散加权成像(diffusion weighted imaging, DWI)稍高信号影(图6)。

1.2 术前评估与手术过程

结合患儿病史、体征及辅助检查,患儿中耳及乳突腔巨大肿物,听骨结构不清,半规管破坏严重,外耳道狭窄。耳道内空间有限,耳内镜下操作困难,耳内切口术式难以彻底清除病灶,增加复发风险。与患儿家属充分沟通病情、术式及分次手术计划后,本次以彻底清除病变为目的,行耳显微镜下右耳乳突改良根治术。于耳后切口,显微镜下开放鼓室和上鼓室,见内部大量胆脂瘤样物质(图7A),膨隆挤压外耳道后顶壁,引起外耳道狭窄。开放乳突向上至鼓室天盖,见胆脂瘤膨隆挤压天盖部分骨质缺失,分离与硬脑膜粘连的上皮,彻底清理。清理胆脂瘤至上鼓室前隐窝及中鼓室面神经膝状神经节处,锤骨头、砧骨体及镫骨板上结构缺失,锤骨柄及鼓膜紧张部结构存在。可见胆脂瘤上皮附着镫骨底板表面及包裹面神经水平段,骨管缺失,清理后见面神经水平段水肿膨出,向后至垂直段部分裸露。乳突深面及向后可见1块畸形骨性隆起(图7B),畸形骨深面可见上皮爬行。为彻底清理上皮,磨除部分畸形骨,随后整块松动并脱离,见片块

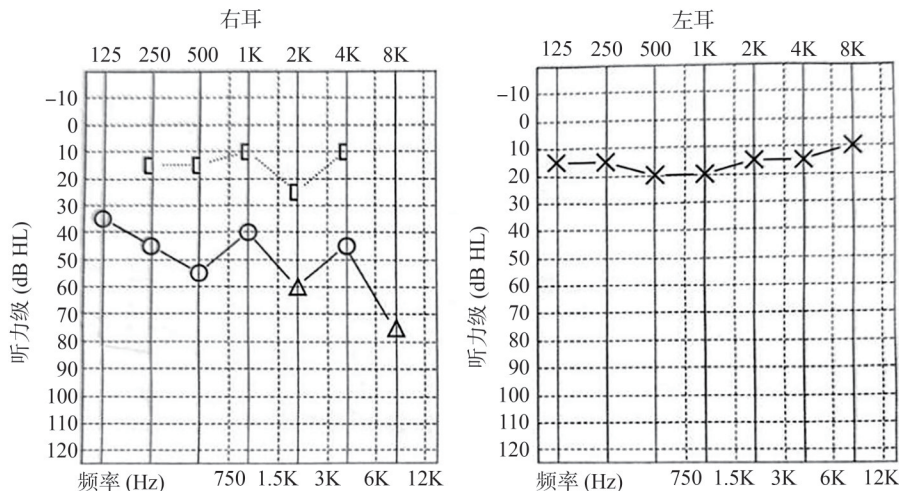


图1 纯音听阈检查

上皮,清理时深面见可疑蓝线,小心剥离上皮后可见蓝线裸露。后半规管骨质部分破坏,外半规管蓝线裸露。用颞肌筋膜覆盖外半规管及面神经裸露处(图7C),行外耳道及耳甲腔成形术。收集骨粉填充后半规管骨质缺如处周围,覆盖筋膜(图7D),取耳甲腔软骨放入中鼓室支撑,形成小鼓室,颞肌筋膜覆盖乳突腔及桥接外耳道皮肤,用明胶海绵堵塞外耳道和乳突,并于外部加压包扎。

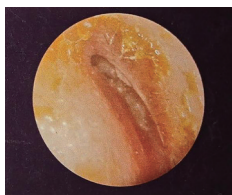


图2 术前耳内镜检查

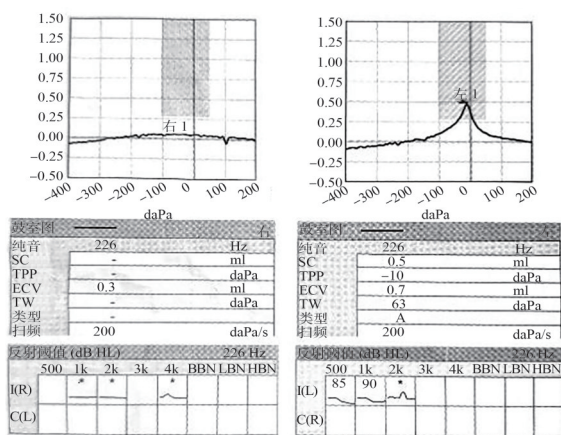


图3 声导抗检查

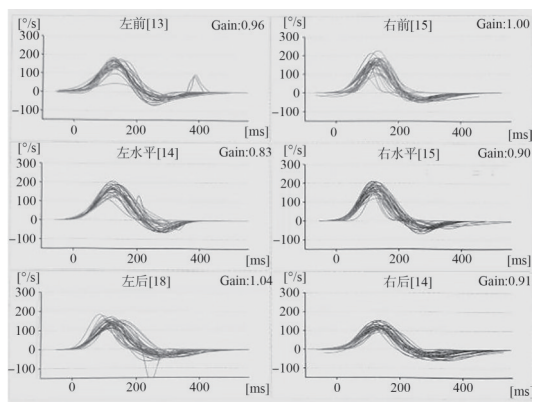


图4 视频头脉冲试验

2 结果

2.1 病理

术后病理诊断示右乳突新生物符合胆脂瘤改变(病理号:447216),见图8。

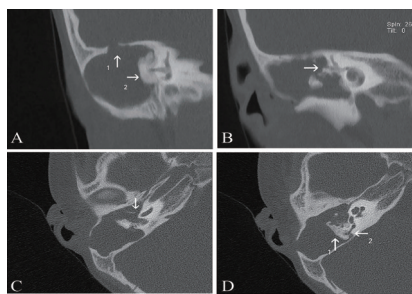


图5 颞骨HRCT

注:A冠状位:1鼓室天盖缺如,2畸形骨块;B冠状位:外半规管缺损;C横断位:锤骨头及砧骨体结构缺如;D横断位:1畸形骨块,2后半规管缺损

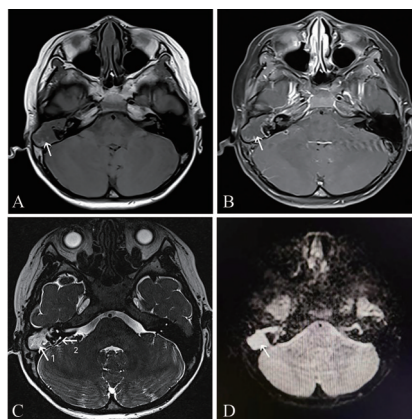


图6 中耳乳突MRI+DWI

注:A横断位T1:乳突内巨大软组织等信号密度影;B横断位T1增强:软组织密度影周边环形强化;C横断位T2:1高信号软组织密度影,考虑为中耳胆脂瘤,2后半规管缺损;D横断位DWI:稍高信号密度影

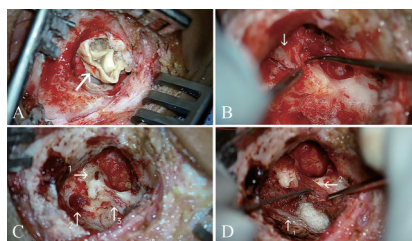


图7 术中所见

注:C1外半规管裂,取筋膜覆盖,2后半规管裂处,取筋膜覆盖,3面神经水平段裸露;D1骨粉填充后半规管骨质缺如周围,2颞肌筋膜覆盖外半规管

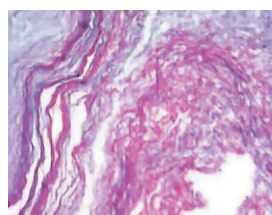


图8 镜下所见

2.2 术后随访

术后1月复查耳内镜见外耳道内少量黄色分泌物,深部黏膜稍潮红肿胀,鼓室潮湿(图9A)。术后2月复查耳内镜示耳道深部上皮化良好,鼓膜完整,无明显穿孔及渗液(图9B)。家属述患儿无耳痛、耳鸣、面瘫、耳流脓以及眩晕、跌倒等不适。

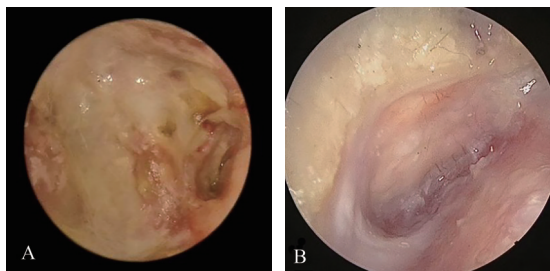


图9 术后复查耳内镜

该患儿目前仍持续随访中,生活质量良好,能正常融入日常学习及生活,无跌倒、头晕及耳鸣等不适。其听力较术前稍差,但能正常沟通交流。患儿家属暂无2次手术计划。嘱其定期复查随访。

3 讨论

CC的发病机制有多种假说,目前广泛接受的为表皮样结构学说。表皮样结构为胎儿中耳发育过程中的一种暂时性外胚层结构,通常在妊娠33周时自行退化消失,若该结构未能在胚胎期正常退化,且持续存在并异常增生,可能演变成CC^[4,5]。先天性耳畸形(congenital ear malformation, CEM)为常见的出生缺陷,我国单纯性CEM的发生率为69.9%,其中中耳畸形占18.6%。中耳腔起源于胚胎早期的第一咽囊(第一鳃弓与第二鳃弓之间的内胚层凹陷),锤骨和砧骨来自第一鳃弓的Meckel软骨,镫骨来自第二鳃弓的Reichert软骨,所有中耳结构均在妊娠第30周完成发育^[6]。CC和CEM的发病机制皆可追溯至胚胎期外胚层组织的发育异常,为其在临床诊疗中的共存提供了理论依据。

根据2017年欧洲耳科与神经耳科学会和日本耳科学会关于中耳胆脂瘤的定义、分类和分期联合共识(European academy of otology and neurotology/Japanese otological society joint consensus statements on the definitions, classification and staging of middle ear cholesteatoma, EAONO/JOS Joint Consensus Statements), CC的标准化诊断前提为无鼓膜穿孔、耳漏病史,但共识说明既往有中耳炎或耳漏病史不能排除CC存在^[7-9]。本病例中患儿外耳道皮肤完整,鼓膜紧张部存在,锤骨柄结

构存在,锤骨头及砧骨体、镫骨上结构缺失。但术中乳突腔深面可见块状游离畸形骨,其深面上皮铺行。考虑锤砧骨融合畸形,同时砧骨镫骨并外半规管发育异常,故考虑先天中耳胆脂瘤合并中耳畸形^[10,11]。出现鼓膜松弛部穿孔、耳漏考虑为继发感染的表现。受咽鼓管解剖结构及免疫力低下导致反复上呼吸道感染影响,儿童胆脂瘤的感染性与侵袭性较成人更严重^[12,13]。胆脂瘤囊性核心不断生长增大,对周围机械性压迫加重,使中耳腔及外界压力失衡,引流受阻,黏膜渗出,中耳微环境改变。因CC发病隐匿性较强,未得到及时诊断及干预,在持续炎症刺激及胆脂瘤分泌酶类的协同作用下鼓膜遭到破坏,继而穿孔,出现反复耳痛、耳漏,标志着疾病从局限性静止状态向侵袭性破坏状态转变^[14,15]。秦兆冰等^[16]报告的3例中耳畸形合并CC患者中2例为锤骨及砧骨融合和镫骨破坏、部分缺失,分别为16岁女性及35岁女性,有鼓膜穿孔、传导性听力损失和耳漏病史,提示并非所有CC的临床表现局限于共识中的标准。本例患儿肿物巨大且病情隐匿,故存在长期炎症刺激导致耳道流脓的表现。

针对CC临床Potsic分期系统,本患儿就诊时为Potsic IV期,乳突受累提示疾病残留的风险增加^[17]。为保护现有结构组织及功能,CC需尽早进行手术治疗。在儿童患者中,内窥镜相较于显微镜技术具有损伤少、恢复快、舒适度高优点,但考虑该患儿年龄偏小且胆脂瘤在乳突腔膨大性生长,侵蚀范围较大。结合上述检查结果,耳道内有效操作空间受限,在狭窄骨性通道内难以完成精细剥离与充分灌洗,无法保证彻底清除病变,且本患儿伴有半规管瘘及面神经裸露,需显微镜下充分磨骨。故优先选用显微镜下乳突开放术^[18]彻底清除胆脂瘤,尽可能多保留耳道骨质,维持原有的解剖和生理状态,以利于重建,同时保留无病变的中耳黏膜结构,为术后中耳形成含气的鼓室腔和维持中耳稳态创造条件^[19]。本次手术存在淋巴液外漏、面神经水肿裸露情况,若强行分离并尝试一期植入人工听骨,不仅面临移植体缺乏稳定支撑基底的解剖学障碍,更将延长手术时间,增加内耳感染及听力损失的风险。在中耳CC的治疗上,多数学者主张患者术后应坚持随访,当出现听力下降或怀疑复发时再考虑2期手术^[20,21]。本案例患者术前已与家属进行充分沟通,明确告知分期手术策略,1期以彻底清除病变为主要目标,术后经6~12个月随访确认无复发,乳突腔上皮化稳定后,再依据听力及影像学结果评估是否进行2期手术。

HRCT对判断胆脂瘤病变范围、周围组织破坏程度以及结构畸形有很大作用^[22],一般作为首选检查方法^[23]。本病例CT显示,右鼓室及乳突窦骨质破坏,累及骨性外耳道后壁并致局部狭窄,术中证实胆脂瘤上皮呈膨胀性

生长,挤压外耳道后骨壁,致其明显内移。膨胀性压迫的影像特征与胆脂瘤上皮通过分泌角蛋白和炎症介质激活破骨细胞、而非肿瘤性侵袭的机制一致。术中见外耳道皮肤尚完整,亦印证了CT所示狭窄为外部压迫所致,为选择外耳道成形手术提供依据。术中发现砧骨、锤骨头畸形融合成骨块可证实先天中耳畸形的存在,结合影像学锤砧关节形态异常、关节间隙消失,呈骨性融合影,确认了CC的诊断,排除后天继发性可能。HRCT提示天盖缺损在术中被证实为硬脑膜暴露并伴有胆脂瘤上皮粘连,但MRI增强扫描未见脑膜异常强化,提示硬脑膜虽受侵犯但尚未发生炎症性增厚或脑膜反应,此对照为术者判断硬脑膜处理的边界提供关键依据。对于发现传导性耳聋、难以察觉的面瘫或反复耳漏、常规治疗效果不佳时,可考虑行HRCT排查中耳畸形及胆脂瘤病变。儿童接受单次CT扫描的健康风险没有明显提高^[24]。但单纯CT对软组织性质分辨能力有限,鉴别胆脂瘤条件不足,不具备特异性,通常建议联合MRI诊断。Fitzek等^[25]2002年首次提出将DWI应用于诊断中耳胆脂瘤,证实其对检测先天性中耳胆脂瘤具有较高的敏感性及特异性,可实现对先天性中耳胆脂瘤的精确诊断^[26],同时在辨别面神经及迷路受累、颅内并发症等方面有帮助。DWI序列的应用可避免不必要的2期手术^[27]。目前,CT、MRI影像融合技术也在逐步推广^[28],结合HRCT及MRI图像可显著提高诊断的正确度,具有优势互补作用,更有利于术前定位^[29,30]。

4 结论

先天性中耳胆脂瘤合并中耳畸形较为隐匿且罕见,在具有传导性耳聋、反复耳漏难以治愈等临床表现的儿童患者身上应尽早完善影像学检查排除该病变。通过对该病例的分析,为基层临床医生提供诊疗经验,提升专科能力,避免漏诊及误诊,并结合病情制定适宜的手术方案,尽可能减轻患儿的痛苦和家庭负担。

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2026年本刊可直接使用缩略语的常用词汇

为节约版面,本刊对常用的英文缩略语词汇作如下规定:以下词汇在正文中(摘要除外)首次出现时写出中文全称和英文缩略语即可,不再要求写出英文全称,再次出现时可直接使用缩略语。

非综合征性听力损失(nonsyndromic hearing loss, NSHL)
感音神经性听力损失(sensorineural hearing loss, SNHL)
听神经瘤(acoustic neuroma, AN)
大前庭水管综合征(large vestibular aqueduct syndrome, LVAS)
分泌性中耳炎(secretory otitis media, SOM)
听性脑干反应(auditory brainstem response, ABR)
电诱发听性脑干反应(electrically evoked auditory brainstem response, EABR)
耳蜗微音电位(cochlear microphonic, CM)
听觉事件相关电位(auditory event related potential, AERP)
耳声发射(otoacoustic emission, OAE)
畸变产物耳声发射(distortion-product otoacoustic emission, DPOAE)
瞬态声诱发耳声发射(transient evoked otoacoustic emission, TEOAE)
自发性耳声发射(spontaneous otoacoustic emission, SOAE)
耳蜗电图(electrocochleography, ECochG)
听觉稳态反应(auditory steady-state response, ASSR)
总和电位(summating potential, SP)
动作电位(action potential, AP)
复合动作电位(compound action potential, CAP)
电诱发复合动作电位(electrically evoked compound action potential, ECAP)
神经反应遥测(neural response telemetry, NRT)
最长声时(maximum phonation time, MPT)
信噪比(signal-to-noise ratio, SNR)
永久性阈移(permanent threshold shift, PTS)
暂时性阈移(temporary threshold shift, TTS)

纯音平均听阈(pure-tone average, PTA)
行为观察测听法(behavioral observation audiometry, BOA)
视觉强化测听法(visual reinforcement audiometry, VRA)
游戏测听法(play audiometry, PA)
患者导向的听觉改善分级(client oriented scale of improvement, COSI)
助听器效果国际调查问卷(international outcome inventory for hearing aids, IOI-HA)
助听器效果评估简表(abbreviated profile of hearing aid benefit, APHAB)
助听器日常使用满意度量表(satisfaction with amplification in daily life, SADL)
噪声下言语感知测试(speech perception in noise, SPIN)
噪声下听力测试(hearing in noise test, HINT)
人工耳蜗(cochlear implant, CI)
内毛细胞(inner hair cell, IHC)
外毛细胞(outer hair cell, OHC)
失匹配负波(mismatch negativity, MMN)
真耳-耦合腔差值(real-ear to coupler difference, RECD)
磁共振成像(magnetic resonance imaging, MRI)
计算机断层成像(computed tomography, CT)
高分辨CT(high resolution computed tomography, HRCT)
言语识别阈(speech recognition threshold, SRT)
聚合酶链反应(polymerase chain reaction, PCR)
声门噪声能量(normalized noise energy, NNE)
自动听性脑干反应(automated auditory brainstem response, AABR)
噪谐比(noise-to-harmonic ratio, NHR)