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胎儿鼻泪管囊肿合并先天性耳聋的偶发病例一例

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摘　要

本病例报告了一名36岁女性患者，因“双侧输卵管堵塞”接受冻胚移植怀孕，孕期检查未见明显异常。停经31周时，产检发现胎儿左侧鼻泪管囊肿，后经羊水穿刺检测发现胎儿有GJB2基因突变，提示可能存在遗传性耳聋。分娩后，胎儿被诊断为双耳轻度感音神经性听力缺失。先天性鼻泪管囊肿通常预后良好，可在出生后自愈，而GJB2基因突变引起的耳聋在遗传性听力障碍中较为常见。患者在孕期并发肺部感染、下肢丹毒及上呼吸道感染，经过适当治疗，胎儿和母亲均顺利度过妊娠晚期。本病例强调了孕期综合管理的重要性，特别是针对具有耳聋家族史的孕妇应加强产前筛查和新生儿听力评估，以便早期发现和干预听力障碍，确保母婴安全。

关键词

鼻泪管囊肿，先天性耳聋，羊水穿刺，产前筛查

An Incidental Case of Fetal Nasolacrimal Duct Cyst Combined with Congenital Deafness

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Abstract

This case report presents a 36-year-old female patient who became pregnant through frozen embryo

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transfer due to “bilateral fallopian tube obstruction”. Prenatal examinations showed no significant abnormalities. At 31 weeks of gestation, an ultrasound revealed a cystic mass in the left side of the fetus’s nasolacrimal duct, and subsequent amniocentesis detected a GJB2 gene mutation, suggesting a potential genetic cause of congenital hearing loss. After delivery, the newborn was diagnosed with bilateral mild sensorineural hearing loss. Congenital nasolacrimal duct cysts generally have a good prognosis and often resolve spontaneously after birth. In contrast, hearing loss caused by GJB2 gene mutations is common among genetic hearing impairments. The patient experienced complications during pregnancy, including pulmonary infection, lower limb erysipelas, and upper respiratory infection, but with appropriate treatment, both the mother and fetus successfully navigated the late stages of pregnancy. This case highlights the importance of comprehensive prenatal management, especially for pregnant women with a family history of hearing loss. It emphasizes the need for enhanced prenatal screening and early newborn hearing assessments to detect and intervene in hearing disorders early, ensuring maternal and fetal safety.

Keywords

Nasolacrimal Duct Cyst, Congenital Deafness, Amniocentesis, Prenatal Screening

1. 临床资料

患者女性，36 岁。因“双侧输卵管堵塞”于外院行胚胎移植术，移植 2 枚冻胚，成活 1 枚。2024-01-13 血清 hCG 为 7153.00 mIU/mL。停经 13 周 6 天行超声检查提示宫内早孕，头臀长 8.4 cm，与孕周相符。孕期规律产检，NT、NIPT、OGTT 及胎儿系统超声检查均未见明显异常。停经 31 周 3 天行产前超声检查，提示胎儿左眼内眦下方可探及一囊性回声，大小约 0.45 cm × 0.17 cm，考虑胎儿左侧鼻泪管囊肿。为进一步明确胎儿情况，孕妇行羊水穿刺检查，过程顺利。染色体微阵列分析(CMA)结果为 arr(X,N)1,(1-22)2，未见染色体数目或结构异常；全外显子测序结果显示 GJB2 基因存在*NM_004004.6:exon2:c.235del (p.L79Cfs3)*变异，判定为致病性变异(PVS1 + PM3_VeryStrong + PS3_Moderate + BS1)，提示胎儿存在遗传性耳聋风险。其后孕期继续随访，未再发现新的胎儿结构异常。停经 39 周 5 天因胎膜早破急诊入院，于 2024 年 9 月 13 日行剖宫产术分娩一活婴，手术顺利。出院诊断包括：胎膜早破、胎儿左侧鼻泪管囊肿、胚胎移植术后、高龄初产等。新生儿出生后行双耳听力筛查未通过。生后 2 月余于外院行听力学检查提示双耳轻度听力异常。随访至 6 月龄，镇静状态下复查听力学检查，结果显示双耳轻度感音神经性听力缺失。追问家族史，患儿祖母存在听力减退病史。

2. 讨论

先天性鼻泪管(nasolacrimal duct, NLD)阻塞被认为是由于胎儿发育过程中未能正常通道化鼻泪管系统[1]。泪道系统在胎儿发育的第 5 周开始形成。在第 10 周，泪道绳索内形成腔道，这与下鼻道腔道的空化同步[2]。通过泪道绳索的通道化，鼻腔下道与泪道之间的通道在第 6 个月的胎儿期开始完成，并持续到足月。如果这一正常的发育过程未能顺利进行，鼻泪管下端可能会形成一层薄的膜状屏障，这种情况在大约 5%~6%的足月新生儿中发生[3]-[5]。NLD 囊肿的胚胎发生归因于鼻泪管系统的哈斯纳膜未能通道化，其出生后患病率为 6%。目前认为，NLD 囊肿的发生主要与胚胎发育异常有关，尚无明确证据表明其具有遗传学基础[6] [7]，多数病例预后良好，可在孕期或出生后短期内自行消退，约 70%以上患儿可在 1 岁

内自愈。

本例中，孕妇因产前超声发现胎儿左侧鼻泪管囊肿而接受羊水穿刺检查，染色体微阵列分析未见异常，但全外显子测序意外发现 GJB2 基因 c.235del 致病性变异，提示胎儿存在遗传性听力损失风险。新生儿出生后听力筛查未通过，后续多次听力学检查证实其存在双耳轻度感音神经性听力缺失，提示产前遗传学结果与出生后表型具有较好的一致性。

遗传性耳聋是先天性听力障碍最常见的原因之一，主要致病基因包括 GJB2、GJB3、SLC26A4 及线粒体 mtDNA，其中 GJB2 突变最为常见，约占遗传性耳聋的 30%~40% [8]。GJB2 基因编码耳蜗内的 Connexin 26 蛋白，其突变可导致耳蜗离子稳态失衡，从而引起感音神经性听力损失，遗传方式多为常染色体隐性遗传[9] [10]。本例中，患儿祖母存在听力减退病史，为遗传背景提供了进一步支持。

需要指出的是，胎儿及新生儿听力损失的病因具有高度异质性，除遗传因素外，宫内感染亦是重要的致病原因之一，尤其是 TORCH 感染中的巨细胞病毒(cytomegalovirus, CMV)，可导致先天性感音神经性听力损失，且部分患儿在出生时缺乏明显的临床表现，仅表现为迟发性或进行性听力下降[11] [12]。本例孕妇孕期曾出现多次感染相关临床情况，因此感染因素，特别是 CMV 感染，在理论上仍需纳入鉴别诊断范围。然而，由于本例未获得明确的产前或新生儿期 TORCH (尤其 CMV)实验室检测结果，感染性听力损失尚不能被完全排除。综合遗传学检测结果、家族史及患儿听力学表现，本研究更倾向于认为遗传因素在本例听力异常的发生中起主要作用。

综上所述，本例为胎儿鼻泪管囊肿合并遗传性耳聋的偶发联合发现。该病例提示，在产前发现胎儿结构异常时，即便该异常本身通常预后良好，亦可作为进一步开展遗传学评估的契机；同时，对于存在遗传性耳聋风险或孕期感染史的孕妇，应加强产前遗传咨询及产后早期听力筛查，以实现听力障碍的早期识别和干预，从而改善患儿的语言及认知发育预后。

声　明

该病例报道已获得病人的知情同意。

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