First Report of Multiple Branchial Cleft Anomalies in Conjunction with a Congenital Dermal Fistula of the Lower Extremity

Ryan Winters, M.D.¹; J. Lindhe Guarisco, M.D.²
¹Tulane University Department of Otolaryngology - Head & Neck Surgery, ²Ochsner Clinic Foundation Department of Otolaryngology - Head & Neck Surgery

CASE REPORT

A 9 year old Caucasian female presented for evaluation of a draining punctum of the skin of the right anterior neck. This lesion had been present for her entire life, and had always had a scant mucoid discharge. For 3 days prior to her presentation she had increased foul-smelling purulent drainage accompanied by sore throat and neck pain. Physical examination was remarkable for an enlarged, medianized right tonsil with peritonsillar abscess, as well as a punctum of the right preauricular skin. This preauricular punctum was non-tender, and scant mucoid fluid was expressible with palpation. Both parents noted that her preauricular area would become tender and edematous with upper respiratory infections (URI), occasionally developing a palpable, cordlike subcutaneous lesion extending toward her external ear. Examination of the ears revealed a 10% posterior superior perforation of the tympanic membrane. No ototomea was present, and the family denied a history of recurrent otitis or otomea associated with URI. The remainder of a head and neck examination was unremarkable. Examination of the extremities revealed a punctum on the dorsum of the right foot, where mucoid fluid could be expressed with palpation. The family reported she had a similar lesion on the posterior right calf. These lesions had become periodically infected in the past, with development of erythema and edema extending down the lateral leg to the foot, accompanied by purulent drainage. This calf lesion had been incised and drained in the past, but the infection had recurred since drainage. Developmentally, she reached all of her milestones appropriately, and was consistently in the 40th percentile for height, weight and head circumference.

Her past medical history was remarkable for multiple other congenital anomalies including atrial septal and ventricular septal defects repaired in infancy, as well as congenital renal anomalies requiring right nephrectomy and bilateral ureteral implantation. She was born at 32 weeks gestation due to maternal preclampsia, and she was noted to have a 2-vessel umbilical cord at delivery.

Computed tomography (CT) of the head and neck with contrast was performed, which revealed an infected sinus tract consistent with a 2nd branchial cleft fistula extending from the right tonsillar fossa to the right anterior neck. A subcutaneous cystic lesion, 1cm in greatest dimension, was noted in the right preauricular area, with no evidence of extension or sinus tract to the ear. The patient underwent successful abscess tonsillecotomy and excision of the infected 2nd branchial cleft fistula.

Intraoperative fistulogram with sialography confirmed the path of the 2nd branchial cleft fistula extending from the tonsillar fossa to the skin of the neck. Intraoperative fistulogram of the 1st branchial cleft cyst, first with methylene blue, then with gastrograffin, demonstrated no fistula to the middle ear and confirmed this as a 1st branchial cleft cyst.

The patient did well postoperatively and has had no recurrence of the 2nd branchial cleft fistula, nor recurrent infections of either the 1st branchial cleft cyst or the peripheral cutaneous fistula of the leg. Medical genetics workup has yielded a normal chromosomal analysis, acetylcholine and carnitine profiles, and no evidence of chromosome 22q11.2. No recognized syndrome has been documented, and the patient appears to have multiple congenital anomalies in association with a single umbilical artery. Orthopedic evaluation of the lower extremities was otherwise unremarkable, and the patient does not suffer any limitations to the use of her right foot.

REFERENCES


CONCLUSIONS

We present the first case of concurrent existence of a 1st branchial cleft cyst, a 2nd branchial cutaneous fistula and a peripheral dermal fistula of the lower extremity. No known syndrome was present in this patient, although multiple congenital anomalies were present on the right side of the body. This represents only the second documented case of such a peripheral dermal sinus. Unfortunately, development of such extremity fistulae remains an area of speculation, and this case, like the only prior documented case before it, cannot conclusively comment on the origin.