CASE REPORT

Lymphatic malformation with acquired Horner syndrome in an infant

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ABSTRACT

An infant presented with right upper eyelid ptosis and was subsequently diagnosed with acquired Horner syndrome. Further evaluation revealed a right-sided cervicothoracic lymphatic malformation. At 13 weeks of age, the child underwent percutaneous intracystic sclerotherapy with a mixture of sodium tetradecyl sulphate and ethanol. Twenty-one weeks after initial treatment, ophthalmic examination showed complete resolution of the blepharoptosis and pupillary miosis. Percutaneous sclerotherapy not only effectively treated the space-occupying lymphatic malformation but also reversed the Horner syndrome that was presumably induced by neural tension (more likely) or compression.

BACKGROUND

Congenital blepharoptosis most commonly results from dystrophy of the levator palpebrae superioris.

Much less commonly it is due to congenital Horner syndrome, a unilateral failure of development in the sympathetic innervation of the eyelid that leads to the triad of blepharoptosis, pupillary miosis, and facial cutaneous anhidrosis, with characteristic heterochromia iridum.¹ Less common still is infantile acquired Horner syndrome, most often attributed to birth trauma, but also associated with neuroblastoma, rhabdomyosarcoma, and Ewing sarcoma.²

The international incidence of lymphatic malformation is about 1 in 6000 to 16 000 live births.³ Of these, 75% occur primarily in the head and neck. Expanding lesions in this anatomical location can disturb third-order sympathetic neurons with resulting ipsilateral Horner syndrome. Surprisingly, there has been no previous report of acquired Horner syndrome resulting from cervicothoracic (or other) lymphatic malformation. This report describes such a case, which when treated with endovascular sclerosis, resolved entirely.

CASE PRESENTATION

An infant presented with 4 days of progressive right upper eyelid droop. The patient's mother described a routine spontaneous vaginal delivery after an unremarkable 41 week gestation without medical illness. Physical examination confirmed unilateral blepharoptosis with ipsilateral pupillary miosis, and anisocoria most evident in darkness. In ambient light, the right pupil measured 3 mm compared with 5 mm on the left (figure 1A). Of note, the irides were of identical blue color. Neuroblastoma was suspected, but urine homovanillic and vanillyl mandelic acid were normal, as were serum levels of neuron-specific enolase and low-density lipoproteins.

Ultrasonography of the neck and chest showed cysts of various sizes with highly echogenic contents located dorsal to the carotid artery and jugular vein. The cysts ranged from less than 10 mm to 20 mm in diameter and no intercystic connection was appreciated. Doppler studies demonstrated no flow within the lesion. Lymphatic malformation was suspected.

Nuclear MRI of the neck and thorax was performed. T2-weighted images revealed extensive hyperintense signal with individual cyst differentiation in the perivascular and posterior cervical regions, beginning at the level of the fourth cervical vertebra, and extending caudally between the sternocleidomastoid and trapezius muscles onto the posterior mediastinum at the level of the second thoracic vertebra (figure 1B). Contralaterally, small paravertebral cysts were identified. After the

Figure 1 Preinterventional (A) photograph showing Horner syndrome of the right eye and (B) fat-saturated coronal T2-weighted MR fast spin-echo image (88/3500) displaying the multicystic lymphatic malformation with high signal intensity.

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Figure 2 Contrast cystogram of the macrocyst before treatment with sodium tetradecyl sulphate and ethanol.

administration of a gadolinium-based contrast agent, homogeneous enhancement of the mass was observed. Combined microcystic and macrocystic lymphatic malformation was confirmed. After consultation with a pediatric surgeon and thorough discussion of management alternatives with the parents, sclerotherapy was planned.

TREATMENT

Ultrasound-guided puncture of a dorsal cyst was performed in the perivascular area using a 5-French coaxial pigtail catheter. Iodine contrast agent was injected, and fluoroscopy confirmed the extent of the lesion with percolation of contrast dye throughout the perivascular lesion as well as in the paravertebral area on the right (figure 2). Hemorrhagic fluid was aspirated from the cysts via the solitary puncture, and 60% ethanol was instilled. After 20 minutes, the blood-stained ethanol was aspirated, and a mixture of sodium tetradecyl sulphate (STS) 3% and lipiodol was injected. A 5-French pigtail drain was inserted with internal and external fixation. CT of the upper thoracic aperture was conducted to confirm the location of the sclerosants within the lymphatic malformation. The drainage system was removed after 3 days.

OUTCOME AND FOLLOW-UP

Repeat MRI was performed at 1 week, 4 weeks, 7 weeks, and 21 weeks (figure 3B) after initial treatment, and resolving thrombosis was noted. Physical examination showed gradual reduction in the size of the lesion beginning at post-treatment week 4 and was imperceptible by post-treatment week 21 (figure 3A). The blepharoptosis appeared intermittently until 16 weeks

Figure 3 Post-treatment (A) photograph obtained 21 weeks after intervention shows complete regression of the Horner syndrome and (B) coronal T2-weighted MR fast spin-echo image (123/7310) showing significant reduction in size.

post-treatment, at which time it resolved entirely. The pupils appeared nearly symmetric after week 21.

DISCUSSION

Lymphatic malformations in some cases can masquerade as tumors, particularly when accompanied by neurological impairment. Lymphatic malformations of the head and neck typically present with signs and symptoms perinatally. Cervical lymphatic malformations can occasionally produce compression of the trachea and esophagus. No previous example has produced Horner syndrome. Interestingly, in the present case, the lesion was large enough to stretch or otherwise perturb the sympathetic trunk, but did not cause aerodigestive compression. An interesting clue to the acquired nature of the Horner syndrome was the absence of heterochromia iridum that is seen with congenital absence of sympathetic stimulation during iris development. Fortunately, treatment in this case was performed promptly because of the risk to the sympathetic nerves, and indeed the Horner syndrome was reversed.

Several methods have been effective in the management of cervical lymphatic malformations including surgery, laser, observation and sclerotherapy.⁴ Because of the specific anatomical location and risk to critical adjacent structures, the multidisciplinary team elected to defer surgery in this case, and did not see a role for intracystic laser. Several alternative sclerosing protocols were considered.

Sclerotherapy of lymphatic malformations has evolved since it was first trialed with bleomycin in 1977.⁵ Subsequent investigations in search of more effective sclerosants with fewer local and systemic untoward effects have identified a variety of agents including tetracycline (1986), OK 432 (1987), and Ethibloc (1993).^{6–8} Despite reports of successful treatment of cervical lymphatic malformations with OK 432 (Picibanil) or bleomycin, there are reports of allergic reaction including anaphylaxis, shock, and death.⁴ Doxycycline induced neurotoxic Horner syndrome in 4 of 49 treatments of head and neck lymphatic malformations in one study.⁹ The combination of ethanol and STS has been well tolerated with few serious deleterious effects and successful resolution of entire lymphatic malformations in a single treatment session.^{10–11} In the present case, the ultrasound suggested individually loculated cysts, which evidently was not the case, since fusion of most of the cysts, uniform percolation of the injected agents, and nearly complete resolution of the entire lesion was observed.

The beneficial effect of treatment using ethanol–STS in this case was noted 4 weeks after treatment, with full effect requiring 21 weeks. Not surprisingly, the signs of the Horner triad resolved at different rates, possibly due to variable injury along the sympathetic chain, but more likely due to end-organ sensitivity to various degrees of sympathetic stimulation.

Contributors All authors contributed significantly and are in agreement with the article's content.

Competing interests None declared.

Patient consent Detail has been removed from this case description/these case descriptions to ensure anonymity. The editors and reviewers have seen the detailed information available and are satisfied that the information backs up the case the authors are making.

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