Human Pathology: Case Reports (2015) **xx**, xxx–xxx

ELSEVIER

Human PATHOLOGY Case Reports

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Case Report

Intraorbital neuromuscular choristoma adjacent to the optic nerve

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Received 16 July 2015; revised 26 August 2015; accepted 2 September 2015

Abstract Neuromuscular choristoma is a rare tumor that incorporates mature skeletal muscle within fascicles of peripheral nerve. The etiology is poorly understood, yet most present in large nerves of children, with a tight link to post-operative fibromatosis recently appreciated. Herein, we report an exceptional intra-orbital example in a 53-year-old man with optic nerve compression.

1. Introduction

Neuromuscular choristoma (a.k.a. neuromuscular hamartoma, benign triton tumor, and nerve rhabdomyoma) is an extremely rare peripheral nerve tumor with an admixture of mature striated muscle and native neural elements [1]. Up to 40–50 cases have been reported using looser definitions, although some in optic or trigeminal nerves likely represent other entities given the inclusion of smooth muscle and/or fat [2]. Most involve larger nerves, such as brachial plexus or sciatic nerve in children, although smaller nerve and intracranial examples have also been reported. Intra-orbital cases are exceptional, with only two other published cases to our knowledge [3,4]. Herein, we report the third example.

2. Case report

A 53 year old man presented with one-year history of slowly progressive left visual loss. He had intermittent sharp retro-orbital pains and a gradually enlarging left central scotoma. On examination, his best corrected visual acuity was 20/20 OD and 20/400 OS. He had a left relative afferent pupillary defect. Extra-ocular motility was normal. Dilated funduscopic examination revealed left optic atrophy.

MRI of the orbits with and without gadolinium disclosed an enhancing 6 × 6 × 6.8 mm nodular lesion in the left orbital apex abutting the inferior aspect of the optic nerve (Fig. 1A and B). He underwent a subtotal resection of the tumor, which was lateral to and separate from the medial rectus at the level of the orbital apex. It grossly appeared as a soft, non-encapsulated, violaceous-brown lesion, which was distinct from the intact left optic nerve.

Histopathology revealed fascicles containing both mature striated muscle and peripheral nerve elements (Figs. 1 and 2). No optic nerve or CNS parenchyma was seen. Histochemical and immunohistochemical stains revealed S-100 (Fig. 2B) and neurofilament (Fig. 2C) staining of Schwann cells and

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http://dx.doi.org/10.1016/j.ehpc.2015.09.001

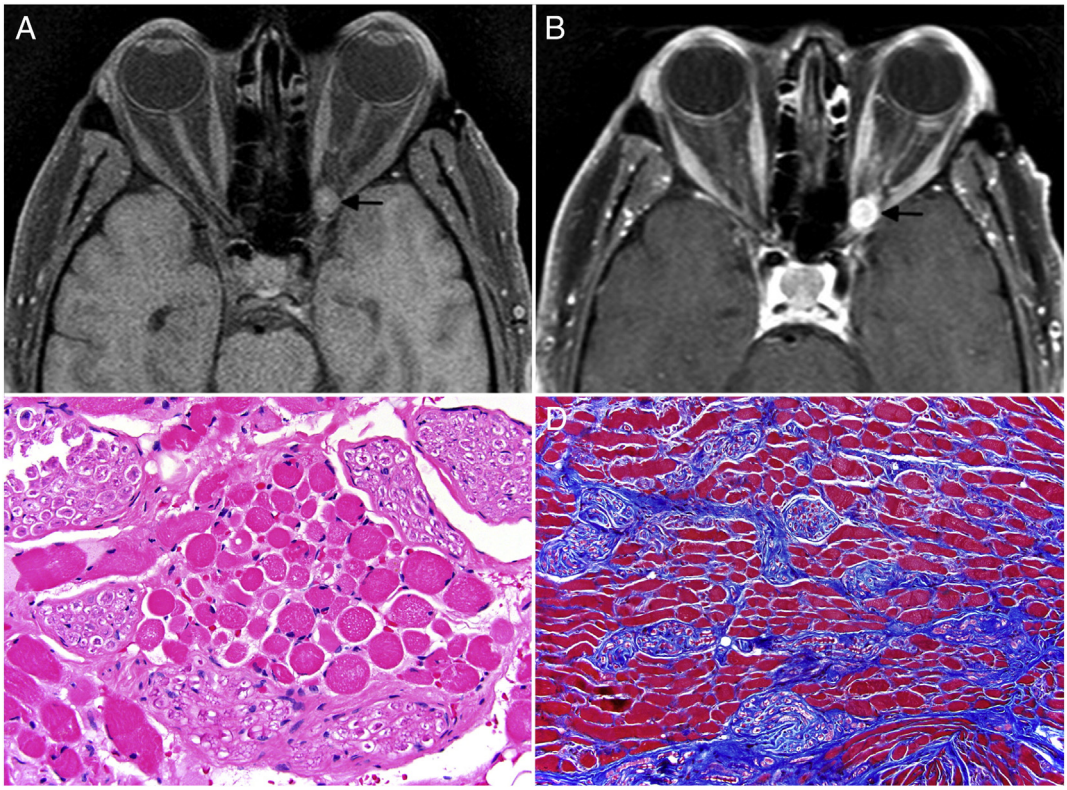

Fig. 1 Fat-saturated T1-weighted axial MRI of the brain without (A) and with (B) gadolinium demonstrates a rounded mass involving the orbital apex adjacent to the left optic nerve. Intra-fascicular admixtures of mature nerve and skeletal muscle were evident on H&E (C; 200×) and trichrome (D; 100×) stains, the latter highlighting muscular elements in red and collagen-rich neural elements in blue.

axons respectively, with individual and grouped neural elements surrounding skeletal muscle fibers. Strong desmin expression was seen in the latter (Fig. 2A). An epithelial membrane antigen stain highlighted perineurium surrounding each nerve fascicle. Perineurium was also seen focally surrounding individual muscle fibers (Fig. 2D). This architectural pattern was consistent with the diagnosis of neuromuscular choristoma.

3. Discussion

Neuromuscular choristoma is a poorly understood, but benign intraneural tumor variably considered malformative (choristoma or hamartoma, possibly due to muscle entrapment during neural development or overgrowth of intraneural muscle spindles), metaplastic (transformation of neuroectodermal elements into skeletal muscle), or neoplastic (rhabdomyoma of nerve) in nature. They have been variably reported to regress, remain stable, or recur following surgery, with recent data suggesting that most of the "recurrences" actually represent aggressive fibromatosis (desmoid), detectable as low-density areas on MR imaging [2]. Whereas most examples have involved large nerves in children, cranial nerve examples have also been reported, albeit more commonly in adults. To our knowledge, this is only the third intra-orbital case and it likely arose from a branch of the oculomotor nerve, with associated compression and atrophy of adjacent optic nerve; nevertheless, the precise origin was not obvious intraoperatively and the normal ocular motility suggests that it did not involve cranial nerve III proper. Prior intra-orbital examples have similarly involved the apex, with reported origins either being unclear or from the oculomotor nerve [3,4]. Familiarity with this rare diagnostic entity can help avoid overly aggressive surgery.

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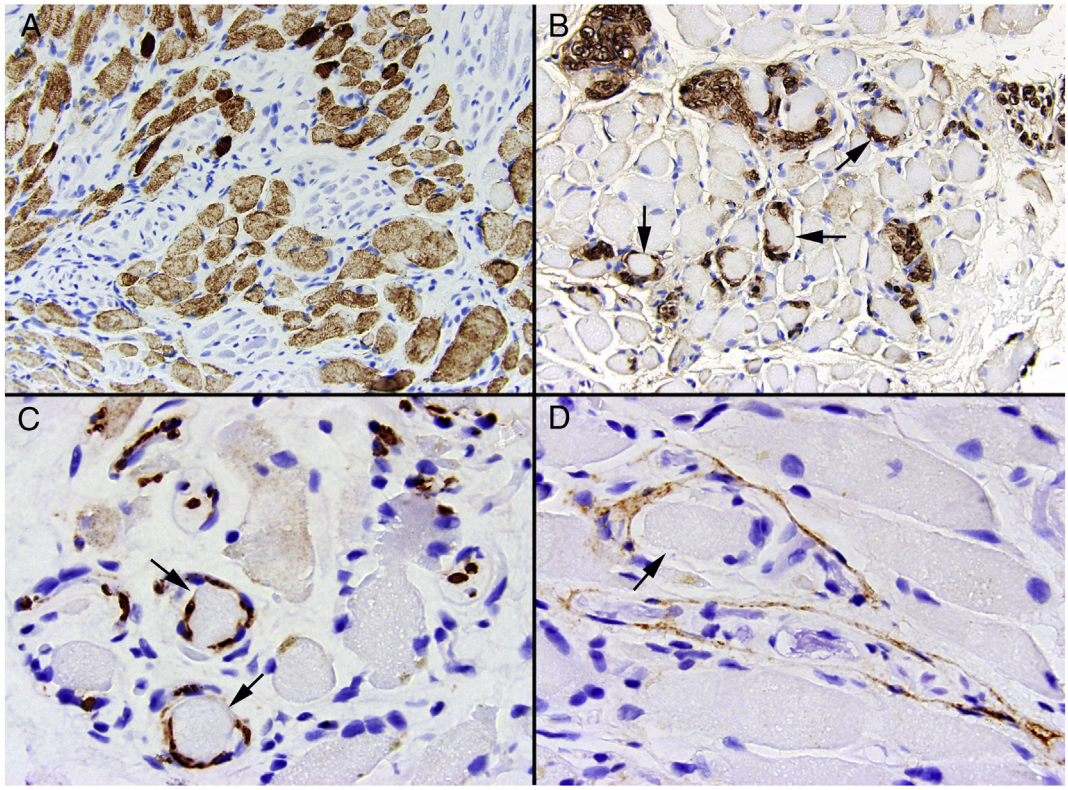

Fig. 2 Immunohistochemistry revealed that the striated muscle fibers were desmin positive (A; 200×), whereas neural elements included S100 positive Schwann cells (B; 200×), neurofilament positive axons (C; 400×), and epithelial membrane antigen positive perineurium (D; 400×). Neural elements often surrounded individual immunonegative muscle fibers (arrows).