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IMAGE – OPEN ACCESS

Neurofibromatosis

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A 25-year-old male presented with progressive swelling on the left side of the ocular orbit ongoing for 2 years that had led to difficulty opening the associated eye for 3 months. There was no history of similar swelling in the family. Examination revealed a firm, irregular, diffuse, pigmented area of swelling on the left side of the orbit extending to the upper and lower eyelids (Fig. 1). There were no café au lait spots, axillary freckles, other subcutaneous swelling, or Lisch nodules present. The results of a biopsy were consistent with neurofibroma (Fig. 2). Surgical excision of the mass was offered, but the patient declined. Genetic counseling was performed.

A neurofibroma is a benign nerve sheath tumor of the peripheral nervous system that originates from non-myelinating Schwann cells. Neurofibromas are most commonly seen in the autosomal-dominant genetic disorder Neurofibromatosis type I, also known as Von Recklinghausen disease (1, 2). Most lesions are asymptomatic; however, pain, itching, and cosmetic disfigurement can occur. Surgical excision of the mass can be performed; however, recurrence is common. Plexiform neurofibromas have a 9% to 12% risk of malignant transformation and warrant aggressive treatment and close follow up (2, 3).

Informed Consent: Written informed consent was obtained from patient who participated in this study.

Peer-review: Externally peer-reviewed.

Conflict of Interest: The author have no conflict of interest to declare.

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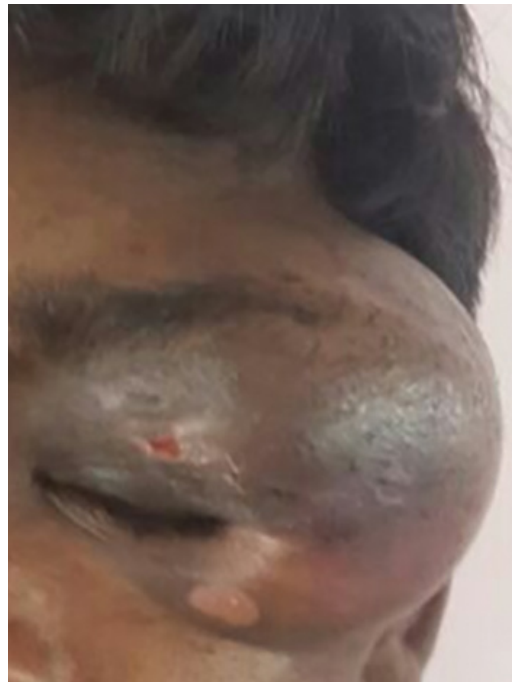

Figure 1. Diffuse, pigmented swelling on the left side of the ocular orbit

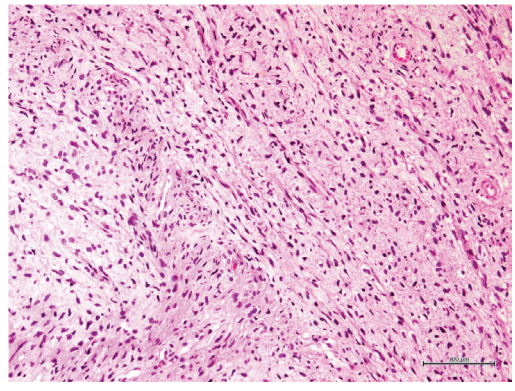

Figure 2. Neurofibroma with spindle cells exhibiting wavy nuclei and fibroblasts in a loose myxoid matrix containing collagen bundles (hematoxylin and eosin; scale bar: 100 μm)

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