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Case Report

A Rare Presentation of Plexiform Neurofibroma

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Abstract

We report on a rare case of a child with persistent right upper lid eversion with conjunctival prolapse since birth that failed various attempts in repositioning the right superior fornix at other centre. He was found to have a right supero-temporal orbital mass above the prolapsed area. Computerized tomography (CT) scan of orbit confirmed a right lacrimal gland tumour with thinning of the right lesser wing of sphenoid. An excision biopsy of the tumour via anterior orbitotomy and eyelid reconstruction were performed. Histopathology report reviewed plexiform neurofibroma of the lacrimal gland. Further physical examination confirmed presence of multiple café-au-lait spots. He was diagnosed as Neurofibromatosis Type 1.

Keywords: Conjunctiva, eyelids, lacrimal gland, neurofibromatosis, plexiform neurofibroma

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Introduction

Plexiform neurofibroma is a hamartoma of neuroectodermal origin. It is a rare tumour that represents 1-2% of orbital tumours (1). It is associated with Neurofibromatosis Type 1, which is inherited in an autosomal dominant manner (2,3). This lesion can involve any peripheral nerve but often involves sensory nerve of orbit or eyelid (1). Peripheral nerve sheath tumours contributes to 4% of all orbital tumours. Neurofibromatosis involving lacrimal gland was reported to be 20.6% (4). To the best of our knowledge, there was no reported case of neurofibroma involving lacrimal gland in a child as young as our patient. Congenital upper lid eversion caused by this lesion was not reported before.

Case Report

A 4-month-old baby boy was referred to our centre for congenital right upper eyelid eversion. This child was born full term, via vaginal delivery, with uneventful antenatal and perinatal history. He was found to have right upper eyelid eversion with conjunctival prolapse since birth (Fig. 1). Both eyes were normal, with clear cornea, deep anterior chamber and clear lens. His fundus examination was normal with pink optic discs, cup-disc-ratio of 0.2 and no choroidal folds. He was otherwise active and well with no syndromic features. We also noted that he had multiple café-au-lait spots on his back and thigh (>6 counts and > 15mm in diameter). There is no family history of malignancy or any known hereditable disease.

Initial computerized tomography scan (CT) of orbit done in the previous referring centre was reported as normal with no intra-orbital mass. He underwent few examinations under anaesthesia with few attempts to reposition his right upper lid but failed in previous referring centre. His right eyelid eversion continued to progress with worsening conjunctival prolapse. Upon examination, a firm, non-tender mass was palpable at supero-temporal region of right orbit, causing complete mechanical ptosis. There was no right eye discharge or surrounding tissue inflammation.

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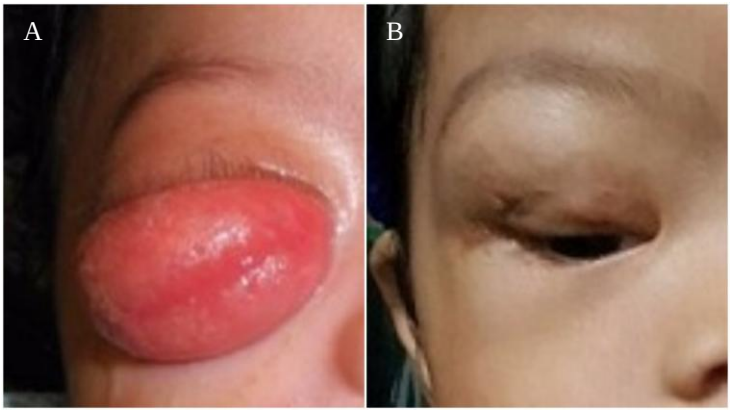

Figure 1: Picture of this 4-month-old child on presentation, showing right upper conjunctival prolapse with upper lid fullness; A) Picture of patient 2 months after the surgery, showing right eye ptosis with resolution of right upper conjunctival prolapse; B) Frontalis suspension surgery was planned for this child.

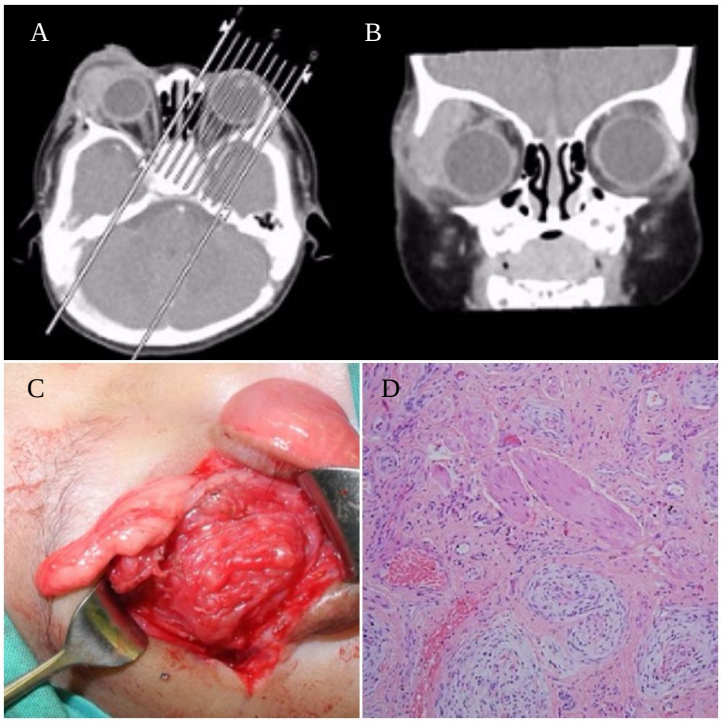

Figure 2: A) CT orbit in axial view (on the left), B) coronal view (middle) showing a well- defined, homogenous mass originating from the right lacrimal gland. The right globe is intact. C) Image of the tumour infiltrating the right lacrimal gland. D) Histopathological image of the tumour specimen, showing irregular unencapsulated tumour composed of rounded to irregular lobules of tumour cells separated by fibrous bands. These tumour cells exhibit oval to spindle shaped cells with variable amount of cytoplasm and indistinct cell border, lie loosely within a myxoid stroma.

A contrast enhanced CT scan of the orbit and brain with smaller cuts revealed a homogenous right lacrimal gland mass (Fig. 2). There was thinning of right lesser wing of sphenoid. He underwent a right orbital mass excision via anterior orbitotomy (Fig. 2) and repositioning of right upper fornix and upper lid reconstruction. His abnormally stretched eyelid was then being trimmed and reconstructed. The dehisced lateral canthus was tightened.

Histopathology report of the excised mass showed plexiform neurofibroma of the lacrimal gland (Fig. 2). He was diagnosed with Neurofibromatosis Type 1. He was referred to paediatrician for further management. On follow-up at 2 months, his right upper eyelid remained ptotic due to prolonged stretching of levator aponeurosis (Fig. 1). He was planned for a right frontalis suspension surgery.

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Discussion

Lacrimal gland mass in children is rare, reported to be 1.8% to 2.4% among all orbital masses in children (5). Lacrimal gland enlargement can be caused by infection, infiltration by benign tissue or malignant tissue and hyperplasia or hypertrophy of structures within the lacrimal gland such as nerve fibres, blood vessels, ductal epithelium, myoepithelium or stromal cells. Plexiform neurofibroma involving lacrimal gland is uncommon, although reported to be 20.6%, (4) lid deformity primarily caused by lacrimal gland mass was rarely reported. Cases that were reported often had involvement of skin, soft tissue or skeletal muscle. Orbito-temporal neurofibromatosis may develop in utero, rapid growth usually occurs during childhood and puberty due to increase in the number and size of neurofibromas. Plexiform neurofibroma may be quiescent until onset of puberty (6).

There were only 4 reported cases of lacrimal gland neurofibroma. There were two adults, a 43-year-old woman and a 63-year-old man. The 43-year-old woman presented with left eye lateral swelling and ptosis. She was initially diagnosed with pan sinusitis but 6 months later, lacrimal gland tumour was confirmed by a CT scan. A biopsy was performed and shown nonspecific fibrosis and inflammation. The lesion did not respond to oral and local injection of steroids. As the tumour was increasing in size over 9 months, excision was done via lateral orbitotomy and her symptoms resolved. Histopathology of the tumour showed benign peripheral nerve sheath tumour compatible with schwannoma or neurofibroma. The 63-year-old man presented with right facial mass and upper lid swelling that gradually increased over 3 years. CT scan confirmed a mass at the right lacrimal gland and at parotid gland. Both right lacrimal gland mass and right parotid gland mass were excised in different setting. The histopathology of lacrimal gland was reported as myxoid neurofibroma. The histopathology of the parotid gland was a well differentiated mucoepidermoid carcinoma (7).

There were 2 children with reported lacrimal gland plexiform neurofibroma, an 8-year-old and a 2-year-old girls. The 8-year-old girl presented with left supero-temporal bluish swelling for 1 year with no other complaints. CT scan revealed a mass extending from orbital margin to temporal fossa. It became painful in the following year, an excision biopsy was performed and histopathology showed plexiform neurofibroma (8). The youngest reported case was a 2-year-old girl who presented with right eye upper lid ptosis since age of 3-4 months. She was seen by an ophthalmologist at 1-year-old for worsening ptosis and S-shaped deformity of right upper eyelid. Magnetic resonance imaging revealed lacrimal gland mass. The mass was excised 6 months later. Histopathology findings were consistent with plexiform neurofibroma (9).

Our patient is the youngest ever reported to have neurofibroma involving the lacrimal gland. Eyelid eversion with conjunctival prolapse as a presentation for this tumour was also not being reported before. Congenital eyelid eversion can be caused by vertical shortening of the anterior lamella of eyelid, vertical elongation of the posterior lamella of eyelid, lateral eyelid elongation, lateral canthal tendon laxity, orbicularis oculi hypotony or failure of orbital septum to fuse with the levator aponeurosis (10). In this case, eyelid eversion could be due to elongation of the eyelid by the lacrimal mass, inability of the orbital septum to fuse with levator aponeurosis and absence of a strong lateral canthal tendon. There was no reported case of eyelid eversion caused by eyelid mass.

As in this case, by excising the neurofibromatosis-infiltrated lacrimal gland with eyelid reconstruction, the prolapsed conjunctiva was repositioned. Post-operatively, he had residual ptosis secondary to levator aponeurosis dehiscence. Frontalis suspension surgery was planned for this child. Early surgery is warranted to prevent development of amblyopia.

Conclusion

Eyelid eversion and conjunctival prolapse since birth is often temporary and usually resolves with time. Persistent eyelid eversion with conjunctival prolapse that did not respond to tarsorrhaphy or did not reduce with time should be investigated for underlying growing mass. Treatment for this type of case is aimed at treating the underlying cause with lid reconstruction.

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