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Rare Case Of Neurofibroma Of Eye Lid

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ABSTRACT

Neurofibromatosis type 1 (NF1) is a genetic neurocutaneous disorder with autosomal dominant inheritance, primarily affecting the skin, nervous system, and eyes. Ocular manifestations include optic pathway gliomas, Lisch nodules, and plexiform neurofibromas etc., In the present case, a solitary neurofibroma was seen on the upper eyelid of a 10-year-old male which impacted his vision. Surgical excision and histopathological analysis confirmed a plexiform neurofibroma. Early recognition of NF1-associated eyelid tumors is crucial for preventing vision impairment, and recent imaging advances support improved diagnostic accuracy for NF1's ocular features.

1. CASE REPORT

The patient is a 10-year old male who is the 1st born child of non-consanguineous marriage presented with a gradually enlarging soft, non-tender swelling on left upper eyelid with an appearance similar to that of a 'bag of worms' (Fig. 1). This mass had caused ptosis and progressive vision impairment over the past 2-3 years. Ophthalmic evaluation revealed that the visual acuity of the left eye was 6/18 with correction. The child was fully immunized and developmentally normal with no similar complaints in the family. Physical examination showed multiple hyper-pigmented patches (cafe au lait spots) and small fibromas distributed over back, abdomen and upper limbs (Fig. 2-4).

Fig 1: Pictorial representation of the left eye-lid evincing neurofibroma

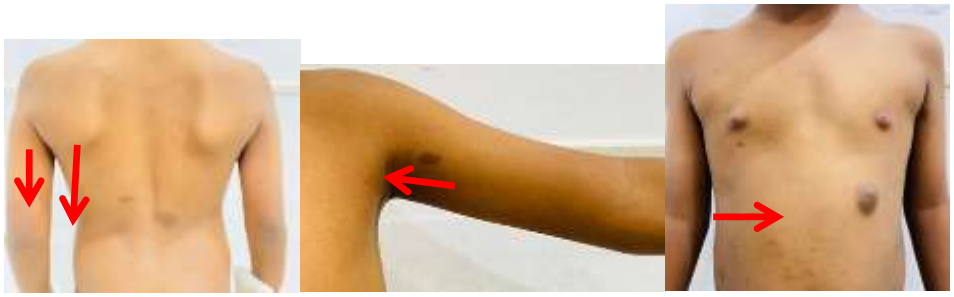


Fig 2-4: Pictorial representation of the hyper-pigmented patches (cafe au lait spots) and small fibromas seen on the back, abdomen and upper limbs

On carrying out MRI BRAIN WITH ORBIT, a well-defined lobulated enhancing soft tissue lesion involving left orbit superior and lateral extraconal compartment with involvement of fat and extraconal muscles was seen.(Fig-5) No involvement of optic nerve was revealed. Multiple focal hyper intensities in the bilateral deep cerebellar white matter, dentate nucleus, middle, superior cerebellar peduncles, dorsal aspect of pons and bilateral cerebral peduncles and posterior limb of internal capsules are suggestive of focal areas of signal intensity (FASI) . MRI findings were suggestive of Neurofibromatosis 1.

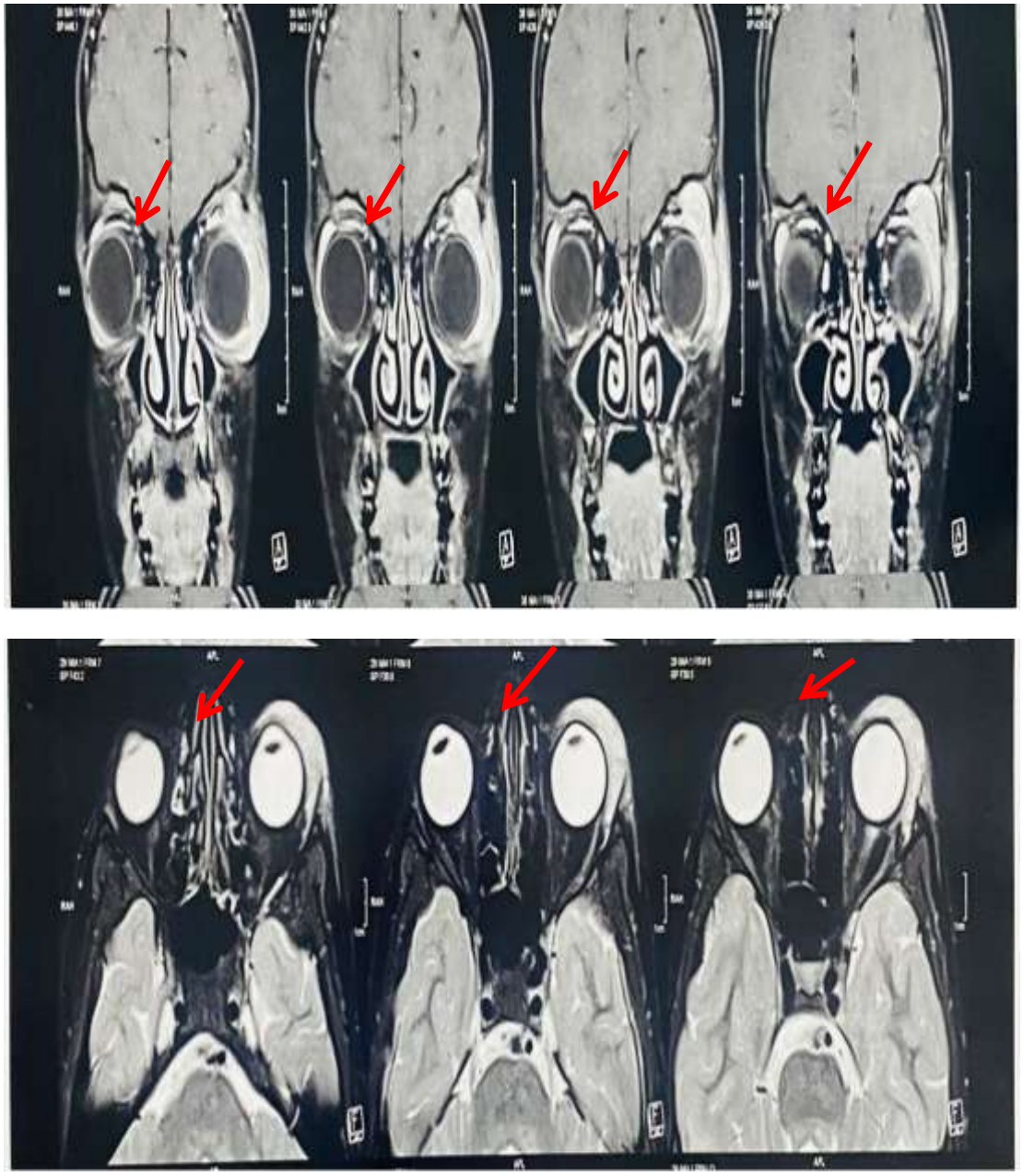
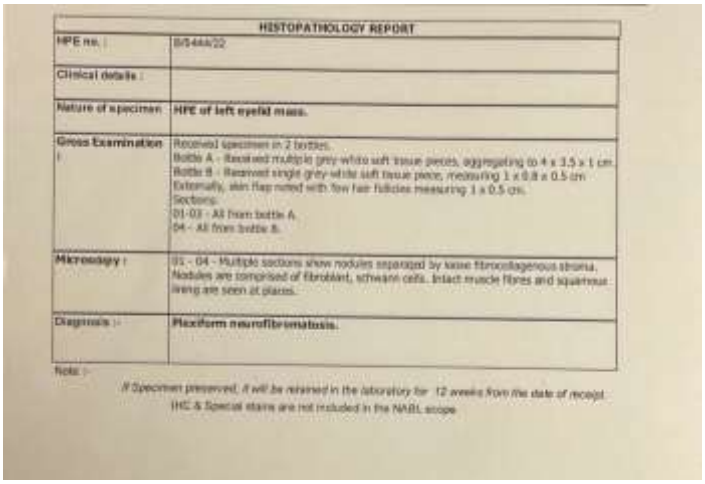


Fig 5 a well-defined lobulated enhancing soft tissue lesion involving left orbit superior and lateral extraconal compartment with involvement of fat and extraconal muscles was seen.

Based on the MRI findings, the patient underwent debulking of tumor with frontalis sling surgery to improve eyelid function. A histopathological examination of the removed mass was ordered. The examination revealed multiple nodules comprising of fibroblast and schwann cells separated by loose fibrocollagenous stroma (Fig. 7). Intact muscle fibers and squamous lining were also seen at places suggestive of plexiform neurofibroma.



HISTOPATHOLOGY REPORT

HPE no. :	B/544/22
Clinical details :	
Nature of specimen :	**HPE of left eyelid mass.**
Gross Examination :	Received specimen in 2 bottles. Bottle A - Received multiple grey white soft tissue pieces, aggregating to 4 x 3.5 x 1 cm. Bottle B - Received single grey white soft tissue piece, measuring 1 x 0.8 x 0.5 cm. Externally, skin flap noted with few hair follicles measuring 1 x 0.5 cm. Sections: 01-03 - All from bottle A. 04 - All from bottle B.
Microscopy :	01 - 04 - Multiple sections show nodules separated by loose fibrocollagenous stroma. Nodules are comprised of fibroblast, schwann cells. Intact muscle fibres and squamous lining are seen at places.
Diagnosis :	**Plexiform neurofibromatosis.**

Note :-

If Specimen preserved, it will be retained in the laboratory for 12 weeks from the date of receipt.

IHC & Special stains are not included in the NABL scope.

Fig 7: Extract of the histopathological report

On post-operative day 5, an improvement in the vision of the left eye (6/9 without correction) was seen.

2. DISCUSSION

NF-1 also known as von Recklinghausen syndrome is a neurocutaneous disorder resulting from the mutation of neurofibromin, a tumor suppressor gene on long arm of chromosome 17 which encodes neurofibromin, a tumor suppressor protein which plays a role in cell signaling. The Neurofibromin 1 gene is a negative regulator of the RAS oncogene signal transduction pathway, controlling cell growth and differentiation. It stimulates the GTPase activity of RAS. It shows greater affinity for RAS p21 protein activator 1, but lowers specific activity.

NF-1 is a disease in the RASopathy family of diseases, which also include diseases such as Costello syndrome, Noonan syndrome, and Cardiofaciocutaneous syndrome.[1] The National Institutes of Health in 1987 had described diagnostic criteria for NF-1. To diagnose NF-1 two or more of the following features should be present – Café au lait macules (CALM) on the skin, iris Lisch nodules, optic pathway glioma, freckles in the axilla or groin area, neurofibroma, neurofibroma, bony dysplasia, and a first-degree relative with NF-1.[2] Solitary neurofibromas have been reported in unusual sites such as tongue, peritoneum, mandible, and palatine tonsil.[3]There are three main types of neurofibromas: localized (most common), diffuse, and plexiform. Although the majority of neurofibromas occur sporadically and have an extremely low risk of malignant transformation, the plexiform type is pathognomonic for neurofibromatosis type 1 (NF 1). It carries an increased risk of malignant transformation.[4]

NF-1 can cause bony dysplasia leading to spinal scoliosis and/or kyphosis, cortical thining of cortex leading to pseudoarthrosis, neurobehavioural developmental disorder such as ADHD (attention deficit hyperactivity syndrome), speech and motor delay, motor deficit, spatial deficit, precocious puberty.There is increase risk of pheochromocytoma, rhabdomyosarcoma, leukemia and wilms tumor compared to general population. [5]

PNFs, which arise from peripheral nerve sheath tissues, are hallmark features of NF1 and occur in approximately 30%-50% of affected individuals. These are benign yet infiltrative tumors are typically congenital but may grow during childhood and adolescence. They frequently affect the head and neck, leading to functional impairment and cosmetic concerns. Orbital PNFs, as in this case, often present with mechanical ptosis, canthal dystrophy, and visual disturbances due to tumor mass effect.Management of PNFs is complex, as surgical excision is often incomplete due to their diffuse growth pattern and proximity to critical structures. In this case, surgical debulking and frontalis sling surgery improved both functional and aesthetic outcomes. However, recurrence is a well-documented challenge in these cases.

The mutant gene is transmitted with an autosomal dominant inheritance with variable expression. Up to 50% of NF-1 cases arise due to spontaneous mutation. Prenatal testing may be used to identify the existence of NF-1 in the fetus. Chorionic venous sampling or amniocentesis can be used to detect NF-1 in the fetus. Selumetinib is a drug approved by FDA in April 2020 for the treatment of NF-1 in pediatric population at 2-3 years of age. It is a MAPK kinase 1 and 2 inhibitor and is indicated for use in pediatric patients who are symptomatic and have plexiform neurofibromas which cannot be operated.[6]

3. CONCLUSION

This case underscores the importance of early identification and a multidisciplinary approach are vital in NF1 management. Comprehensive care should address potential complications, such as malignancy transformation (occurring in 5%-10% of PNFs), neurodevelopmental delays, and psychosocial challenges. Regular monitoring with imaging and ophthalmologic assessments is crucial to mitigate disease progression and optimize patient quality of life.

REFERENCES

[1] Wallace, MR; Marchuk, DA; Andersen, LB; Letcher, R; Odeh, HM; Saulino, AM; Fountain, JW; Brereton, A; Nicholson, J; Mitchell, AL (13 July 1990). "Type 1 neurofibromatosis gene: identification of a large transcript disrupted in three NF1 patients". Science.

[2] Neurofibromatosis. . Conference statement. National Institutes of Health consensus development conference Arch Neurol. 1988;45:575–8

[3] Chen N, Hsu YH, Lee YC. Solitary neurofibroma of eyelid masquerading as chalazion Int Med Case Rep J. 2017;10:177–9

[4] Lassmann H, Jurecka W, Lassmann G, Gebhart W, Matras H, Watzek G. Different types of benign nerve sheath tumors. Light microscopy, electron microscopy and autoradiography. Virchows Arch A Pathol Anat Histol. 1977 Sep 28;375(3):197-210. [PubMed]

[5] "Neurofibromatosis 1: Current Issues in Diagnosis, Therapy, and Patient Management", by David Viskochil MD PhD, Mountain States Genetic Foundation, Denver.

[6] AstraZeneca Pharmaceuticals LP.Koselugo (Selumetinib) U.S. Food and Drug Administration Accessed June 21, 2020.