

Maxillofacial Radiology 203

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CASE

A 19-year-old male patient, known with a diagnosis of neurofibromatosis type 1, presented with a plexiform neurofibroma involving the left orbit, zygoma, temporal, and parotid regions. The patient reported a history of left eye enucleation 7-years-ago. What are the pertinent radiologic findings? (Figs.1 and 2)



Fig.1: Panoramic radiograph

INTERPRETATION

The panoramic radiograph shows extensive displacement of teeth and bony components on the left side. There is extensive expansion of the lingula, the entry point of the inferior alveolar nerve into the mandible. Additionally, there is expansion of the infratemporal fossa region and inferior

displacement of the left zygomatic arch. An incidental finding of an impacted canine obstructed by a compound odontoma can also be appreciated in the right anterior mandible. Magnetic resonance imaging (MRI) showed an extensive lobulated T₂ / STIR hyperintense mass involving the left orbit, infratemporal fossa, masseter, parotid, carotid and posterior cervical spaces. The lesion demonstrated central areas of hypointensity within the lobules, consistent with a characteristic target sign.

Neurofibromatosis type 1 (NF1) is an autosomal dominant disorder caused by the mutation of the *NF1* gene that encodes for the protein neurofibromin, a tumour suppressor gene. Patients with this condition frequently present with multiple benign peripheral nerve sheath tumors called neurofibromas. A subtype, plexiform neurofibroma (PN), presents clinically with a so-called 'bag of worms' consistency which is essentially pathognomonic of the syndrome and presents with more extensive nerve plexus involvement. PNs are seen in 17% of patients with *NF1* and are frequently diagnosed

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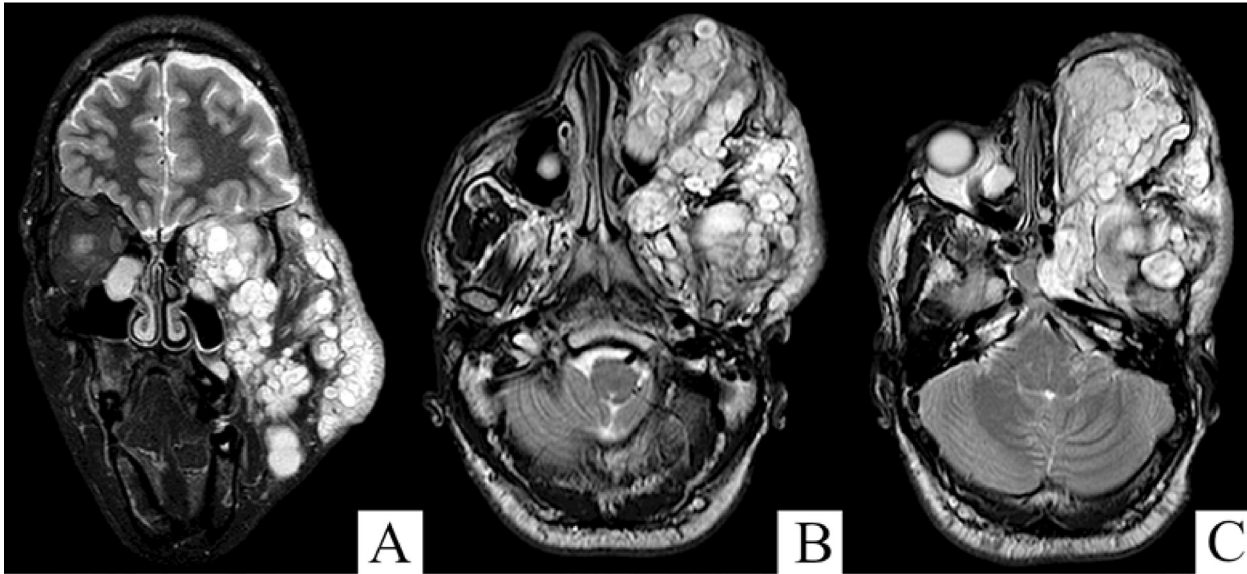


Fig.2: (A) Coronal short-T₁ Inversion Recovery (STIR) and (B,C) axial T₂W MRI images

during the first decade of life.¹ They frequently occur on the trunk (43%), followed by the head and neck region (42%) and the limbs (15%).¹ Patients can often present with multiple plexiform neurofibromas^{1,2,3}, with 42% of head and neck lesions affecting the orbit.¹ Accompanying symptoms are related to mass effect, including loss of function and associated pain in the affected region.¹

Radiologically, PNs present with three growth patterns: not superficial, displacing and invasive. Superficial growth is seen in 25% of cases and is located on the skin or subcutaneously without muscle penetration.² Displacing growth is seen in 26% of cases and presents with a well-defined multinodular appearance that displaces and compresses adjacent structures.² The invasive pattern is seen in 49% of cases and penetrates into joints, muscles and surrounding structures with a multinodular appearance and poorly-defined borders.² The volume of PNs can range between 76ml and 3703ml in size, which may be accurately detected by automated methods on MRI imaging.⁴

The significance of the identification of plexiform neurofibromas includes a potential for malignant transformation (3%), commonly seen in the displacing and invasive growth patterns.^{1,2} Roughly 54% of the invasive growth patterns are seen in the head and neck area.² The management of patients with *NF1* includes family genetic consultation, close clinical follow-up of lesions, and surgical removal of lesions that result in aesthetic or functional impairment.¹ Due to the extensive nature of these lesions, complete surgical resection is not possible in all cases. Median tumour progression is 0.6% per year after surgery, with a higher growth rate noted in young individuals.⁵ The assessment of the lesion size in cases of PN on MRI is advantageous in the monitoring of residual tumours and novel medical treatments.⁴

Authors declaration

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REFERENCES

1. Waggoner DJ, Towbin J, Gottesman G, Gutmann DH. Clinic-based study of plexiform neurofibromas in neurofibromatosis 1. *American journal of medical genetics*. 2000 May 15;92(2):132-5.
2. Mautner VF, Hartmann M, Kluwe L, Friedrich RE, Fünsterer C. MRI growth patterns of plexiform neurofibromas in patients with neurofibromatosis type 1. *Neuroradiology*. 2006 Mar;48(3):160-5.
3. Ehara Y, Koga M, Imafuku S, Yamamoto O, Yoshida Y. Distribution of diffuse plexiform neurofibroma on the body surface in patients with neurofibromatosis 1. *The Journal of Dermatology*. 2020 Feb;47(2):190-2.
4. Solomon J, Warren K, Dombi E, Patronas N, Widemann B. Automated detection and volume measurement of plexiform neurofibromas in neurofibromatosis 1 using magnetic resonance imaging. *Computerized Medical Imaging and Graphics*. 2004 Jul 1;28(5):257-65.
5. Nguyen R, Ibrahim C, Friedrich RE, Westphal M, Schuhmann M, Mautner VF. Growth behavior of plexiform neurofibromas after surgery. *Genetics in Medicine*. 2013 Sep;15(9):691-7