Picture Images in Medicine – Neurofibromatosis

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CLINICAL SCENARIO

A 12-year-old female child reported to the department of paediatrics with a painless swelling of the right side of the hard palate and displaced tooth on the same side [Figures 1a and b]. The swelling was noticed at the age of 6 years and has been increasing in size over the period. There was a positive family history but no other relevant medical history.

On physical examination, he had several café-au'-lait macules and freckling spots on the legs and palms.

A biopsy of the oral swelling revealed densely packed collagen bundles interspersed with wavy bundles of the nerve tissue having spindle nuclei. No dysplastic features were reported [Figures 2a and b].

QUESTIONS

1. What is the most likely diagnosis?
2. List differential diagnosis of this condition?
3. What is the inheritance pattern of this condition?
4. This patient later developed blindness in the right eye. What is your explanation?
5. List 5 features of bony involvement?
6. What is the specific treatment of this clinical condition?

ANSWERS

1. Neurofibromatosis type 1 (NF1 Von Recklinghausen disease).
2. Differential diagnosis
   i. NF2
   ii. McCune-Albright syndrome
   iii. Multiple endocrine neoplasia, type 2b
   iv. Familial multiple café-au'-lait spots.
3. NF1 is considered to have an autosomal dominant pattern of inheritance. People with this condition are born with one mutated copy of the *NF1* gene in each cell. In about half of cases, the altered gene is inherited from an affected parent.
4. Optic gliomas are present in approximately 15% of patients with NF1.
5. Features of bony involvement
   i. Scoliosis
   ii. Short stature
   iii. Sphenoid wing dysplasia
   iv. Cortical thinning
   v. Fractures
   vi. Pseudo-arthrosis.
6. There is no specific treatment for NF; the management includes genetic counselling and early detection of treatable conditions or complications. For this case, surgical excision of the oral neurofibroma will be of help.

NFs are genetic disorders that predispose to tumours formation on nerve tissue. The three main types include NF1 that is usually diagnosed in childhood, NF2 and schwannomatosis. The latter two are usually diagnosed in early adulthood. NF1 is characterised by skin lesions such as brown freckling spots on the trunk (café-au'-lait spots) and axillary freckling; brown pigmentation in the eyes (lisch nodules) and brown dome lesion on the body (benign and malignant nervous system tumours, usually benign neurofibromas). They may also present with bone dysplasia.

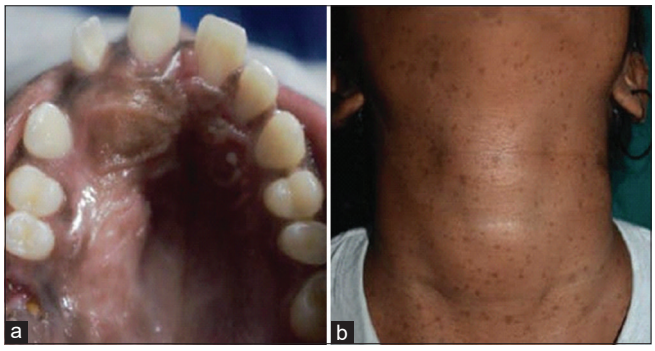

Figure 1: (a and b) Picture of a 12 year old female with a painless swelling of the right side of the hard palate and displaced tooth on the same side

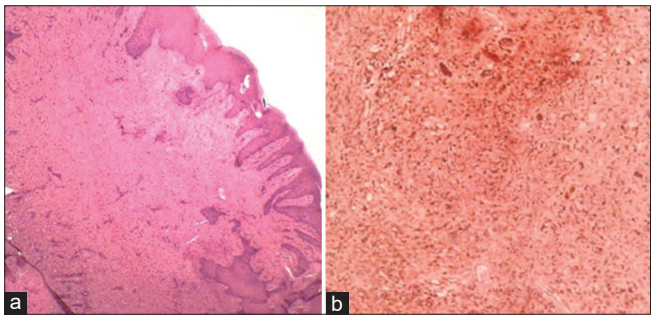

Figure 2: (a and b) Histologic findings of oral swelling showing densely packed collagen bundles interspersed with wavy bundles of the nerve tissue having spindle nuclei

The diagnosis is usually made in a prepubescent child if there are six or more café-au'-lait spots greater than 5 mm in diameter. Diameter of greater than 15 mm is used in adults.

Although there are no specific definitive treatments, patients benefit from genetic counselling, early management of complications, pain control medications, stereotactic surgery to reduce large tumours and irradiation.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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