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Case Report of Neurofibromatosis Type 1

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Abstract **Case Report**

Neurofibromatosis type 1 (NF1) or Von Recklinghausen's disease is a rare genetic disorder which is characterized by the formation of multiple benign tumors arising from the nerves and skin called as neurofibromas and consists of areas of hypopigmentation or hyperpigmentation of skin(café au lait macules). We report a case of 11 years old female child presented with complaints of swelling over forehead, both sides of neck and plantar aspect of foot. On examination multiple café au lait spots were seen. The diagnosis NF-1 was made according to the presence of two or more diagnostic criteria of the National Institute of Health Consensus Development Conference. Neurofibromatosis is an autosomal dominant disease that requires multidisciplinary approach requiring the care from paediatrician, ophthalmologist,dermatologist and advice of genetic counsellor..

Keywords: Neurofibromatosis type 1; Recklinghausen's disease; Café-au-lait macules.

INTRODUCTION

Neurofibromatosis type 1 (NF1) or Von Recklinghausen's disease is a rare genetic disorder which is characterized by the formation of multiple benign tumors arising from the nerves and skin called as neurofibromas and consists of areas of hypopigmentation or hyperpigmentation of skin. Neurofibromatosis type I (NF-1) is the most common type of the disease accounting for 90% of the cases, and is characterized by multiple café-au-lait spots and the development of neurofibromas along peripheral nerves.

CASE REPORT

History

A 11 year old female child presented to Paediatrics outpatient Department by her mother who noticed swelling over forehead, both sides of neck and plantar surface of left foot.

Physical examination

Chocolate brown coloured round to oval macules measuring >0. 5cm noted over trunk and arms –café au lait spots. 4 macules are seen around neck region, 3 posterior and 1 on the forearm.

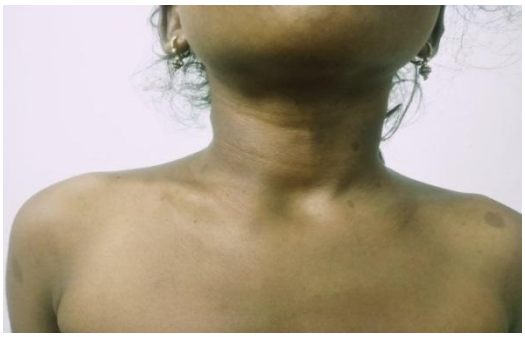

Fig-1: café au lait macules seen over the anterior aspect of neck

Fig-2: Café au lait macule on the forearm

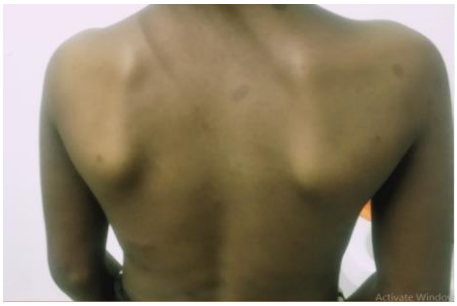

Fig-3: café au lait macules on the posterior surface of chest

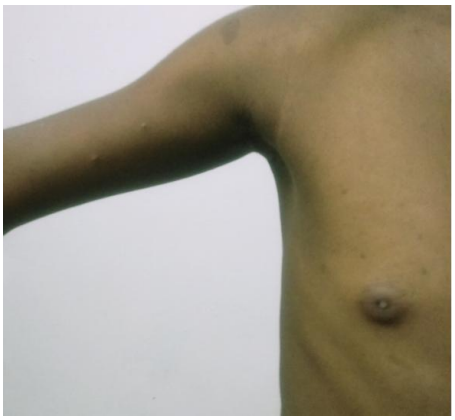

Fig-4: Multiple hyperpigmented macules of size 1-2 mm noted all over body including axilla- crowe's sign positive

Bilateral matted firm swellings palpable along posterior border of sternocleido mastoid.

Small subcutaneous nodules palpable in left interscapular and left lumbar area

A soft non cystic swelling present over the medial aspect of plantar surface of left foot with hyperpigmented skin over swelling- plexiform neurofibroma

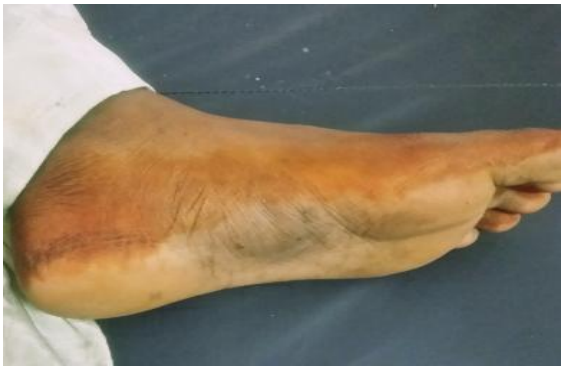

Fig-4: Plexiform neurofibroma on the plantar surface of left foot

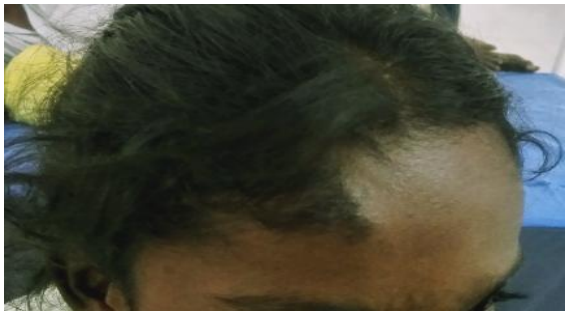

Fig-5: Neurofibroma on the forehead.

A similar swelling found on forehead.

Ophthalmologic examination

Bilateral Lisch nodules- multiple tiny white coloured spots noted on iris of both eyes without visual impairment.

Further evaluation

Routine laboratory investigations were within normal range. X ray and CT brain –normal. Otologic evaluation of 8th nerve has confirmed no damage. Neurologist consultation did not found any alterations in central and peripheral nervous system. Histology confirmed the diagnosis of Neurofibromatosis.

Diagnosis

The diagnosis NF-1 was made according to the presence of two or more of the diagnostic criteria of the National Institute of Health Consensus Development Conference [1]:

- 6 or more café-au-lait macules (>0.5 cm in children of >1.5 cm in adults)
- 2 or more cutaneous or subcutaneous neurofibromas or one plexiform neurofibroma
- Axillary or groin freckling
- Optic glioma
- 2 or more Lisch nodules (iris hamartomas visualized on slit lamp examination)
- Sphenoid wing dysplasia or bowing of long bone (with or without pseudarthrosis)
- First degree relative with an NF1 diagnosis

DISCUSSION

The cases of neurofibromatosis were reported in literature for the first time in XIII century by Madigan, Schaw, and Masello[2]. In the year 1882 - Friederich Daniel Von Recklinghausen recognized NF as the tumors that has their orgination along major peripheral nerves and described that these patients have pigmented skin areas. So, this condition is often referred to as von Recklinghausen's disease [2].

Neurofibromatosis-1 is an autosomal dominant disorder which has an incidence of 1 in 3000 people worldwide [3]. It is characterised by germ-line–inactivating mutation on the Nf1 gene in chromosome 17q11.2 that codes for neurofibromin, a tumor suppressor protein which leads to increased susceptibility to neoplasms of neural crest origin [4].

Neurofibromatosis-1 usually appear in childhood. It consists of 90% of all the cases. They usually present with multiple cafe-au-lait spots and development of neurofibromas along peripheral nerves, Lisch nodules on the iris of eyes and cutaneous neurofibromas. Possible complications in childhood include the development of an optic glioma, endocrine disturbances and involvement of the lower urinary tract. The children may also present with learning disabilities [5].

The patient presented to our department was routinely monitored for complications. The child was annually examined for following conditions

- Assessment of skin to look for new neurofibromas or progression of existing lesions

- Blood pressure monitoring
- Growth and development evaluation
- Eye examination
- Evaluation of skeletal system
- Assessment of learning of the child

CONCLUSION

Neurofibromatosis is an autosomal dominant disease that requires multidisciplinary approach requiring the care from paediatrician, ophthalmologist, dermatologist and advice of genetic counsellor.

Ethical Approval

Necessary approval was taken from the Institution and the patients for carrying out this work

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