National Journal of Clinical Orthopaedics

ISSN (P): 2521-3466
ISSN (E): 2521-3474

www.orthoresearchjournal.com
2022; 6(1): 01-03
Received: 03-10-2021
Accepted: 05-12-2021

Sourav Bansal
Int. GMC, Amritsar, Punjab, India

Dr. Dixit Bansal
Junior Residents, Department of Orthopaedics, GMC, Amritsar, Punjab, India

Aguilera-Alvarez Victor H
MD, MPH Student, Imperial College London, London

Anusha Manoj Kallamvalappil
MBBS, Saveetha Medical College and Hospital, Chennai Tamil Nadu, India

Sharan Kumar Balakrishnan Sundararajan
MBBS, Saveetha Medical College and Hospital, Chennai Tamil Nadu, India

Arghadip Das
Medical Student, Nilratan Sircar Medical College, Kolkata, India

Himani J Suthar
M.B.B.S, Gmers Medical College, Gandhinagar,

Alejandra Lopez
M.D, Mexico City, Universidad La Salle, Mexico

Corresponding Author:
Sourav Bansal
Int. GMC, Amritsar, Punjab, India

Rare case report of neurofibromatosis with distal radial Schwannoma

Sourav Bansal, Dr. Dixit Bansal, Aguilera-Alvarez Victor H, Anusha Manoj Kallamvalappil, Sharan Kumar Balakrishnan Sundararajan, Arghadip Das, Himani J Suthar and Alejandra Lopez

DOI: https://doi.org/10.33545/orthor.2021.v6.i1a.339

Abstract
The term neurofibromatosis (NF) is referred to a group of genetic disorders that primarily affect the cell growth of neural tissues with rare musculoskeletal manifestations. We reported a case of Neurofibromatosis with distal schwannoma in 50 years old female.

Keywords: Cutaneous nodules, Neurofibromatosis, Von Recklinghausen's disease

Introduction
The term neurofibromatosis (NF) is referred to a group of genetic disorders that primarily affect the cell growth of neural tissues. There are two forms of NF: Type 1 (NF1) and type 2 (NF2). These two forms have few common features and are caused by mutations on different genes [1].

Neurofibromatosis type 1, also known as von Recklinghausen's disease, is a neurodermal dysplasia [2]. This disease was first described by Friederich Daniel Von Recklinghausen, the pathologist, in 1882. The pathological alterations behind it begin in the embryonic period, prior to differentiation of the neural crest [3]. NF1 is the most common type of NF and is estimated to occur in 90% of all cases. It is one of the most frequent human genetic diseases, with a prevalence of one in 3,000 births. There is no sex or racial predilection. NF1 is an autosomal dominant disease caused by a spectrum of mutations that affect the *NF1* gene located at the 17q11.2 chromosome [4]. It has one of the highest rates of spontaneous mutation among genetic diseases in humans. Only 50% of the NF1 patients have a positive family history of the disease. The rest of the patients represent spontaneous mutations. The expressivity of the disease is extremely variable, with manifestations ranging from mild lesions to several complications and functional impairment. The penetrance, otherwise, is 100% [5]. However, pathologic changes may also affect other organs and systems, including skeletal system (scoliosis, hypostature, osteoporosis, pseudarthrosis and sphenoid wing dysplasia) or cardiovascular system (hypertension, inherited cardiovascular malformations).

We reported a case of Neurofibromatosis with Musculoskeletal manifestations in 50 years old female.

Case report
A 54 years old female presented with multiple soft tissue cutaneous nodules on the body since childhood. She did not have any systemic problems in the organs of her body or any oral manifestation. All her off springs had skin neurofibroma and Café-au-lait pigmentations in variable sizes and frequencies. She mentioned that her mother and aunt had Café-au-lait spots and neurofibroma on their skin.

A fibrotic nodule was obtained from left arm which was subjected to histopathological examination. Aspirated blood admixed 10 ml fluid showed moderately cellular smears show cluster of plump spindle cells and few group of round to oval cells with centrally placed nuclei and foamy macrophages. Features were suggestive of spindle cell lesions with cystic degeneration possible of schwannoma. CT angiography left forearm showed well defined oval

shaped cystic/ necrotic lesion with mild enhancing irregular wall and septation in antero- lateral aspect of distal forearm.

Discussion

Neurofibromatosis type 1 was first described by Frederich von Recklinghausen in 1882 [6]. Neurofibromatosis type 1 (NF-1) or von Recklinghausen disease is an autosomal dominant disorder with a basic defect in the embryonic neural crest cells which give rise to ectodermal and mesodermal derivatives that affect one in 3000 live births. Oral manifestations of NF have been reported in only 4%–7% of the affected persons [7]. Palatal swellings may occur from a variety of etiological factors and can originate from the structures within the palate or beyond it. They may be painful when secondarily infected. The mean age of occurrence is 27.5 years with a slight predilection toward females. Treatment for both NF-1 and NF-2 is directed toward controlling symptoms and managing the complications [8]. Nallanchakrava *et al* [9] reported a case of a 12-year-old girl with NF-1. The disease started in childhood with the appearance of multiple hyperpigmented skin macules. The girl presents generalized freckling and café au lait spots throughout the body and a diffused swelling measuring about 4 cm × 3 cm, extending from the right maxillary hard palate region to the midpalate. The diagnosis of NF-1 was made according to the presence of two or more diagnostic criteria of the National Institute of Health Consensus Development Conference. No recurrence was observed in a 15-month follow-up after extensive surgical ablation.

Neurofibromas are benign complex tumors. They arise from peripheral nerve sheaths and constitute one of the main manifestations of NF1. A solitary neurofibroma may occur in an individual who does not have NF1, but multiple neurofibromas tend to develop in a person with NF1. Clinical observations suggest that there are at least two major types of neurofibroma which may differ widely in their natural history: 'Discrete' or 'localized' and 'plexiform' neurofibroma [10].

A localized neurofibroma arises from a single site along a peripheral nerve and presents as a focal mass with well-defined margins. It can occur superficially or may involve deeper peripheral nerves [11]. A localized neurofibroma is the most common type of neurofibroma occurring in NF1 patients. They are rarely, if ever, present at birth and usually appear in late childhood or early adolescence. The number of localized neurofibromas tends to increase with age, which varies widely from person to person. NF1 patients may have few, hundreds, or even thousands of localized neurofibromas [12]. Neurofibromas are found mostly on the skin. Nevertheless, many organs may be involved, including the stomach, intestines, kidney, bladder, larynx, and heart. In the head and neck region, the most commonly affected sites are the scalp, cheek, neck, and oral cavity. Several soft tissue lesions, comparable to the localized neurofibroma, could be seen all over the mother's body. Plexiform neurofibroma spreads along the peripheral nerve and may affect some nervous rami [13]. The cranial nerves most involved are the fifth, ninth, and tenth. Abnormal bone development and a deficiency in bone mineral density can cause bone deformities such as a curved spine (scoliosis) or a bowed lower leg. Some children have abnormally formed bones, which can result in bowing of the legs and fractures that sometimes don't heal. NF1 can cause curvature of the spine (scoliosis) that may need bracing or surgery. NF1 is also associated with decreased bone mineral density, which increases the risk of weak bones (osteoporosis). However present case also showed growth on left forearm with osteolytic lesions which can make bone more prone to fracture. So such patients should be warned for such incidences.

Fig 1: Cutaneous nodules on body

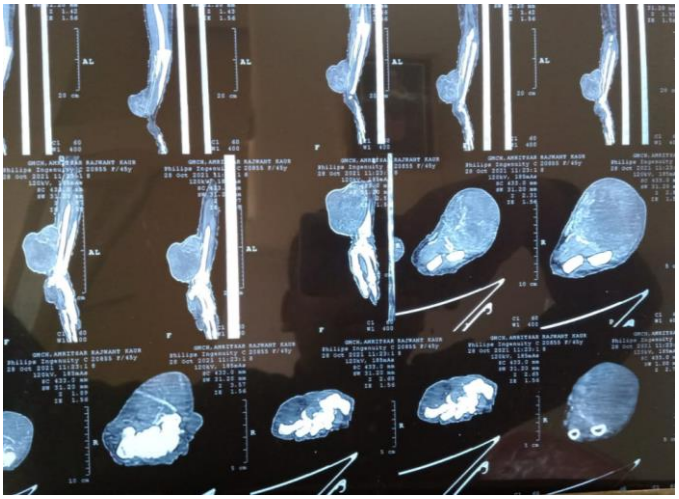

Fig 2: CT scan

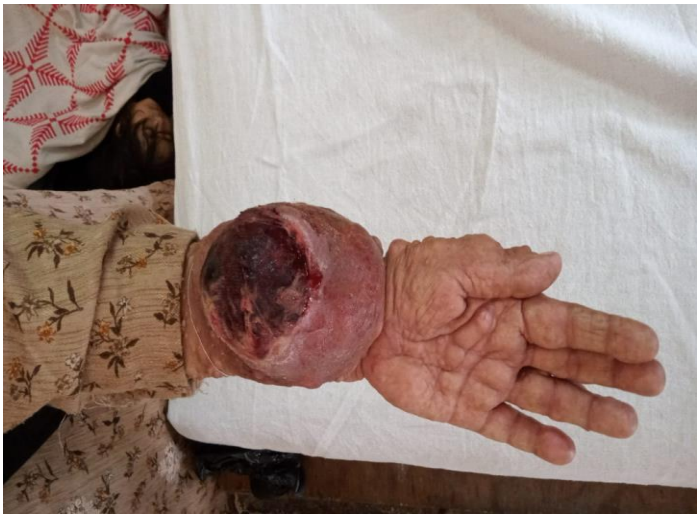

Fig 3: Cutaneous nodules on left forearm

Conclusion

NF is a multisystem disorder requiring management by multiple disciplines, often coordinated through a primary care physician mand a dermatologist. Considering other advanced investigations such as CBCT, immunohistochemical (S-100 protein test) is often beneficial for an accurate diagnosis. Hence, early detection with advanced techniques and investigations with complete resection minimizes the reoccurrence of such tumors.

References

1. Allen CM, Miloro M. Gingival lesion of recent onset in a patient with neurofibromatosis. Oral Surg Oral Med Oral Pathol Oral Radiol Endod. 1997;84:595-7.
2. Barnesn L. Tumours of the nervous system. In: Surgical Pathology of the Head and Neck. 3rd ed. New York: Informa Healthcare USA. 2007, 681-8.
3. Brosius S. A history of von Recklinghausen's NF1. J His Neuro Sci. 2010;19:333-48.
4. Shapiro SD, Abramovitch K, Van Dis ML, Skoczylas LJ, Langlais RP, Jorgenson RJ, *et al*. Neurofibromatosis: Oral and radiographic manifestations. Oral Surg Oral Med Oral Pathol. 1984;58:493-8.
5. Patil S, Khandelwal S, Doni B, Rahman F. A Review of 9 Palatal Swellings. Pak Dent Assoc. 2013;22:134-9.
6. Nafarzadeh SH, Motallebnejad M, Ghorbani A, Jamaatlou N. A case report of neurofibromatosis. Caspian J Dent Res. 2014;3:47-51.
7. Lin IC, Chiu YW, Lui MT, Kao SY, Chen YW. Solitary diffuse neurofibroma of tongue-A case report. Chin J Periodontol. 2012;23:187-96.
8. Vora RA, Mintz DN, Athanasian EA. Intraosseous schwannoma of the metacarpal. Skeletal Radiol. 2000;29:224-6.
9. Nallanchakrava S, Mallela MK, Jeenepalli VS, Niharika HM. A rare case report of neurofibromatosis type 1 in a 12-year-old child: A 15-month follow-up. J Oral Maxillofac Pathol. 2020;24:S106-9.
10. Theos A, Korf BR, American College of Physicians, American Physiological Society. Pathophysiology of neurofibromatosis type 1. Ann Intern Med. 2006;144:842-9.
11. Vivek N, Manikandhan R, James PC, Rajeev R. Solitary intraosseous neurofibroma of mandible. Indian J Dent Res. 2006;17:135-8.
12. Neville BW, Hann J, Narang R, Garen P. Oral neurofibrosarcoma associated with neurofibromatosis type I. Oral Surg Oral Med Oral Pathol. 1991;72:456-61.
13. Needle MN, Cnaan A, Dattilo J, Chatten J, Phillips PC, Shochat S, *et al*. Prognostic signs in the surgical management of plexiform neurofibroma: The Children's Hospital of Philadelphia experience, 1974-1994. J Pediatr. 1997;131:678-82.