Clinical Case Report

Medicine®

OPEN

Lady with wings: a case report of giant neurofibromatosis type I

Suraj Maharjan, MD, Xiucun Li, MD, Jianli Cui, PhD, MD, Yang Liu, MD, Laijin Lu, PhD, MD*

Abstract

Rationale: Neurofibromatosis type I (NF-I) accounts for approximately 90% of neurofibromatosis. NF-I is an autosomal dominant genetic disease which results from the gene mutation of NF-I situated in chromosome 17q11.2.

Patient concerns: A 32-year-old lady presented with a giant wing like structure on her back which started growing from her childhood.

Diagnosis: A diagnosis of NF-I was confirmed as she presented with multiple cutaneous nodules, multiple café-au-lait macules of different sizes, scoliosis deformity, and positive family history of neurofibroma.

Interventions: Surgical excision of tumor and multiple Z plasty reconstruction of the back was carried out.

Outcomes: The excised neurofibroma weighed 6.7 kg containing thickened nerves, nerve roots, and circuitous vessels. The histopathological report confirmed plexiform and diffuse type cutaneous neurofibroma without any malignant transformation. Surgical excision and reconstruction with regular follow-up is an excellent choice of treatment for such a giant neurofibroma as in this case.

Lessons: NF-I is a genetic disease which could present as a giant cutaneous neurofibroma. One of the treatment options for giant neurofibromas causing deformity and physical disability is by surgical excision and histopathological examination with regular follow-up for NF-I recurrence.

Abbreviations: NF-I = neurofibromatosis type I, PNF = plexiform neurofibroma.

Keywords: excision, giant, nerve, neurofibroma

1. Introduction

Neurofibromatosis type I (NF-I) is an autosomal dominant genetic disease which results from the gene mutation of NF-I situated in chromosome 17q11.2.[1,2] NF-I accounts for approximately 90% cases of neurofibromatosis.[3] Although, there are many reports on giant neurofibromas,[4–6] only few of them are complex neurofibromas consisting of plexiform and diffuse cutaneous neurofibromas. We report a case of giant wing like neurofibroma at the back of a 32-year-old lady.

2. Case report

An approval from the ethics committee of the first Hospital of Jilin University was obtained for this case report study. A 32-year-old lady weighing 37.5 kg and 147 cm tall presented with overhanging wing like mass on her back with scoliosis deformity. Initially the mass was a small pea size nodule over the right anterior chest wall which gradually increased in size since early childhood and extended to the back with wings like presentation by adolescence. She has a positive family history of neurofibromatosis. On examination, the mass measured 60 cm × 41 cm with surface ulceration at the distal end. Multiple café-au-lait macules of different sizes and hard cutaneous nodules were present at the back, axilla, and thoracoabdominal regions (Fig. 1A–C). She did not give history of past medical or surgical intervention. All the routine laboratory tests were normal. Thoraco-abdominal plain radiography antero-posterior view showed scoliosis deformity of cervical and thoracic spine.[7] Magnetic resonance imaging revealed "S" shaped curve with an angular deformity at thoracic vertebrae involving T1–T7. No other abnormalities were seen in magnetic resonance imaging regarding centrum, intervertebral disk, and spinal cord (Fig. 2). The patient did not undergo genetic testing. The patient was diagnosed NF-I (plexiform type) from her history and clinical examination.

The tumor excision and reconstruction was done with multiple Z-plasty. The tumor weighed 6.7 kg, containing thickened subcutaneous nerves and nerve roots, large and diffuse circuitous vessels (Fig. 1D, E). Histopathological exam reported giant neurofibroma with components of plexiform and diffuse cutaneous neurofibromas (Fig. 2D, E). At 15 months follow-up, there was complete healing of reconstructed skin at the back with no signs of tumor regrowth. The patient was extremely satisfied with the outcome and ability to carry out her daily activities (Fig. 1F).

3. Discussion

Its diagnosis is based on 2 or more of the diagnostic criteria mentioned in Table 1.[8] Our patient met the criteria for the diagnosis of NF-I due to the presence of the following: positive

Editor: Mauro Alaibac.

The authors have no funding and conflicts of interest to disclose.

Department of Hand and Foot Surgery, The First Hospital of Jilin University, Changchun, Jilin Province, P.R. China.

** Correspondence: Laijin Lu, Department of Hand and Foot Surgery, The First Hospital of Jilin University, Changchun, Jilin Province 130021, P.R. China (e-mail: lulaijin007@yahoo.com).*

Medicine (2017) 96:29(e7523)

Received: 6 March 2017 / Received in final form: 22 June 2017 / Accepted: 23 June 2017

http://dx.doi.org/10.1097/MD.0000000000007523

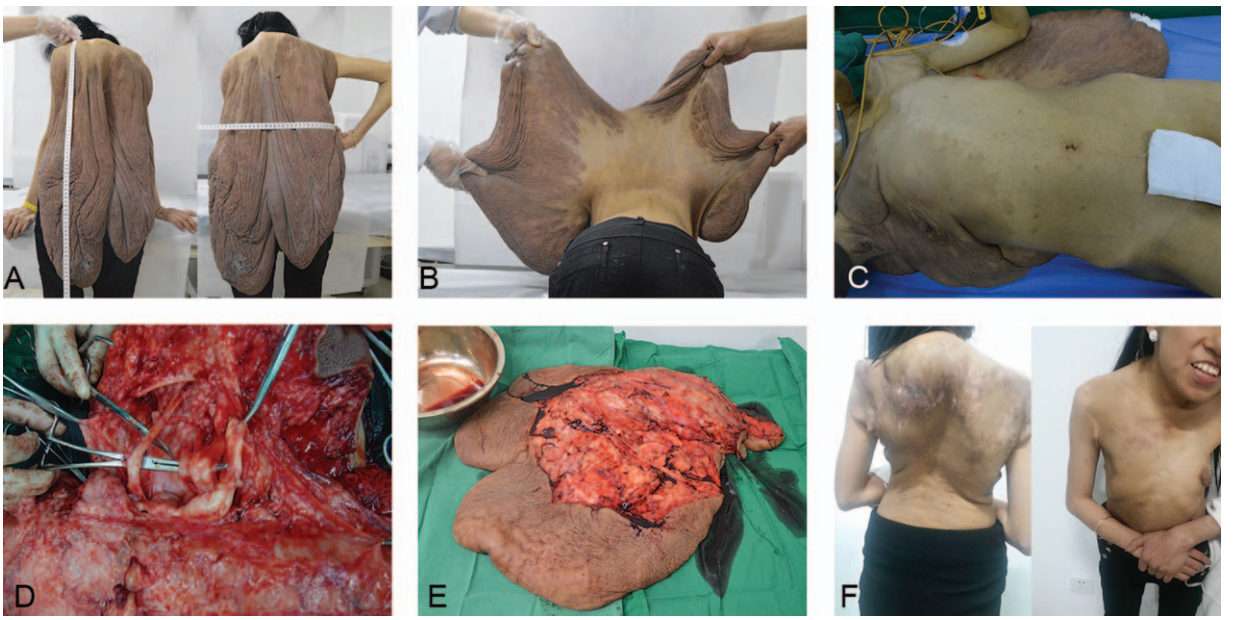

Figure 1. Clinical presentation of the patient. (A, B) Giant wing shaped tumor; (B, C) café-au-lait spots and cutaneous nodules; (D) thickened subcutaneous nerves and nerve roots; (E) excised tumor; and (F) follow-up after 15 months.

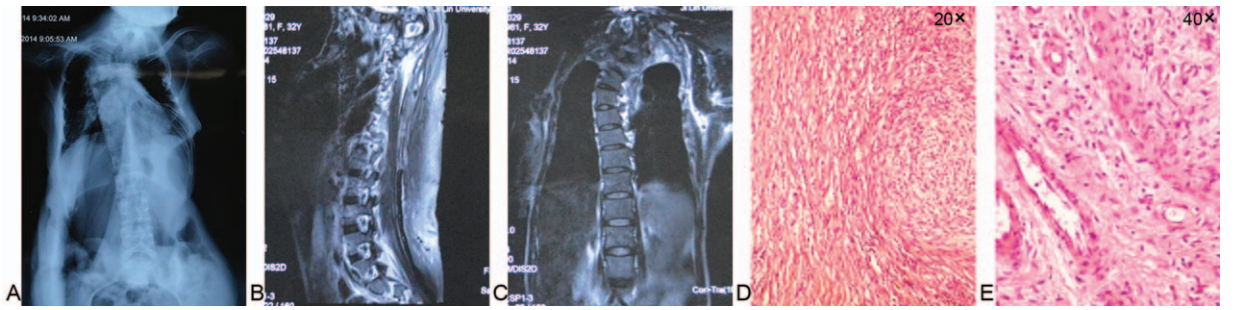

Figure 2. X-ray and magnetic resonance imaging (MRI) spine, and histopathological findings. (A) Anteroposterior thoraco-abdominal X-ray; (B, C) the results of MRI sagittal sections: cervical and thoracic spine scoliosis deformity, and neurofibroma at the back. (D, E) Histopathological findings: cellular matrix containing fibroblasts, Schwann cells, and collagen (hematoxylin and eosin 20× and 40× magnification).

family history of her mother, brother, and son, all diagnosed with NF-I; 6 or more café-au-lait spots in the axilla, back, and thoracoabdominal regions; and multiple cutaneous nodules (Fig. 1A–C).

The initial manifestation of NF-I is usually café-au-lait spots.[9] Nodular neurofibromas can appear in peripheral nerves at any regions of the body and have the capability of growing into a large size,[10] whereas, diffuse cutaneous neurofibromas are more common in adults, ranging from several to many thousands.[1] The neurofibromas over the trunk is one of the common manifestations which accounts for 64% followed by upper extremity (54%), head and neck (48%), and lower extremity (31%).[11] Although, plexiform neurofibromas (PNFs) are analogous to cutaneous neurofibromas, they comprise mainly of extracellular matrix and vessels.[9] They usually are derived from dorsal spinal roots, large nerve trunks, or sympathetic chains.[9] PNF accounts for approximately 30% of patients with NF-I, of which most of them are frequently in the cranio maxillofacial region.[2] Diffuse PNF of the faciocervical region rarely appears after the 1st year of life, and diffuse PNF of other parts of the body rarely develops after adolescence.[1] Furthermore, in approximately 2% to 16% of patients, the nodular and PNFs can transform to malignant peripheral nerve sheath tumors.[10] The tumor in our patient gradually increased in size from chlidhood to adulthood. The weight of tumor excised was 6.7 kg but the histopathological examination did not show any malignant transformation.

With regard to the treatment of these giant tumors, surgical treatment still remains the mainstream, aiming at improving

Table 1

The diagnostic criteria for NF-I[8].

NIH consensus development conference 1988
Six or more café-au-lait macules over 5 mm in greatest diameter in prepubertal individuals and over 15 mm in greatest diameter in postpubertal individuals.
Two or more neurofibromas of any type or 1 PNF.
Freckling in the axillary or inguinal region.
Optic glioma.
Two or more Lisch nodules (iris hamartomas).
A distinctive osseous lesion such as sphenoid dysplasia or thinning of long bone cortex with or without pseudarthrosis.
A 1st-degree relative (parent, sibling, or offspring) with NF-I.

NF-I = neurofibromatosis type I, NIH = National Institutes of Health, PNF = plexiform neurofibroma.

functional and aesthetic effect.[4–6] However, we agree that the most difficult challenge for this surgery is the variation of anatomy and unclear tumor margin. In addition, the tumor contains thickened cord of nerves along with abundant abnormal vessels; therefore, excessive hemorrhage during the operation is also a brainteaser. Furthermore, if the nerve roots of neurofibromas are from major nerves, surgical resection may result in functional impairment. Therefore, the surgeon must cautiously design a detailed preoperative treatment strategy. For the giant neurofibroma of the back of in our patient, resection of the tumor was successfully performed and the defect was covered using local skin flaps with multiple Z-plasty reconstruction. Postoperative wound infection and skin necrosis was not observed. Furthermore, our patient also presented with scoliosis deformity of cervical and thoracic spine,[7] but she refused spinal orthopedic consultation as she had no neurological symptoms and signs. Follow-up at 15 months revealed no sign of tumor recurrence with satisfactory healing. Moreover, shedding the heavy weight from her back has benefited the patient physically as well as psychologically. However, we recommend regular long-term follow-up to assess tumor recurrence. We conclude that surgical excision and reconstruction with regular follow-up is an excellent treatment of choice for giant neurofibromas as in our case.

References

[1] Jett K, Friedman JM. Clinical and genetic aspects of neurofibromatosis 1. Genet Med 2010;12:1–1.
[2] Tchernev G, Chokoeva AA, Patterson JW, et al. Plexiform neurofibroma: a case report. Medicine (Baltimore) 2016;95:e2663.
[3] Gajeski BL, Kettner NW, Awwad EE, et al. Neurofibromatosis type I: clinical and imaging features of Von Recklinghausen's disease. J Manipulative Physiol Ther 2003;26:116–27.
[4] Zhou J, Li M, Luo C, et al. Giant neurofibroma in the right lower limb of a 26-year-old woman: report of a case. Int Surg 2012;97:71–7.
[5] Yuan SM, Cui L, Guo Y, et al. Surgical management of giant neurofibroma in soft tissue: a single-center retrospective analysis. Int J Clin Exp Med 2015;8:5245–53.
[6] Cheng XB, Lei DL, Li YP, et al. Surgical treatment of a giant neurofibroma. J Craniofac Surg 2011;22:2244–6.
[7] Crawford AH, Herrera-Soto J. Scoliosis associated with neurofibromatosis. Orthop Clin North Am 2007;38:553–62.
[8] Neurofibromatosis. Conference statement. National Institutes of Health Consensus Development ConferenceArch Neurol 1988;45:575–8.
[9] Tonsgard JH. Clinical manifestations and management of neurofibromatosis type 1. Semin Pediatr Neurol 2006;13:2–7.
[10] Reynolds RM, Browning GG, Nawroz I, et al. Von Recklinghausen's neurofibromatosis: neurofibromatosis type 1. Lancet 2003;361:1552–4.
[11] Levine SM, Levine E, Taub PJ, et al. Electrosurgical excision technique for the treatment of multiple cutaneous lesions in neurofibromatosis type I. J Plast Reconstr Aesthet Surg 2008;61:958–62.