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Case Report

Neurofibromatosis type 1 manifesting with adolescent idiopathic scoliosis: a case report and literature review

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ABSTRACT

Type 1 neurofibromatosis (NF 1) may present with a constellation of symptoms but literature has recorded that the commonest manifestations are orthopedic symptoms with spinal presentations taking the lead. Of the spinal manifestations, scoliosis is frequently found compared to others which could include spondylolisthesis or defective pedicles and dural ectasia on radiographs. We reported is a 15-year-old girl with NF 1 coexisting with severe thoracolumbar scoliosis. She complains of dull aching pain in her upper back and hips with progressively worsening bending of the back and slight difficulty with breathing. On examination, multiple Café au lait spots on the trunk and legs. No neurological deficits noted. There was no family history of neurofibromatosis. There was severe thoracolumbar scoliosis on X-ray of the low back. The patient then had long segment fusion and stabilization. Bone graft was used to achieve solid arthrodesis. Scoliosis is the commonest manifestation of NF 1 with different severities and degrees of Cobb's angle adolescent females are mostly affected and management is dependent of degree of curvature, comorbidities like NF 1 and type of scoliosis.

Keywords: Neurofibromatosis, Café au lait, Spondylolisthesis, Case report

INTRODUCTION

Neurofibromatosis type 1 (NF 1), also known as the Von Recklinghausen disease is inherited in an autosomal dominant pattern. It is the commonest autosomal dominant disease with a prevalence of about 1:3000. NF 1 gene, the defective gene is located on the long arm of chromosome 17 whose product is Neurofibromin.[1-3] Although a single gene mutation, NF 1 can manifest in multiple organ systems to varying degrees of severity. A diagnostic criterion for NF 1 was first described in 1987. A diagnosis can be made in the presence of two or more parameters of the criteria.[1,4] There are two types of NF as described by literature: type 1 or peripheral NF and type 2 or the central NF. NF 1 is characterized by multiple café-au-lait spots, skinfold (axillary or inguinal) freckling in non–sun exposed areas (lentiginous macules), iris Lisch nodules, tumors of the nervous system, and other features.[5]

Orthopedic involvement is the most common clinical presentation of NF 1 patients with the spinal abnormalities more frequently affected. The incidence of spinal deformities in NF 1 patients varies from 2% to 64% with scoliosis being the most recorded.[1,4] These deformities are traditionally classified into non dystrophic and dystrophic types based on the radiographic evaluation.[6] The non-dystrophic type has the presentations similar to the idiopathic scoliosis, however, the dystrophic type has its

distinctive features, including vertebral scalloping, rib penciling, transverse process spindling, vertebral wedging, paravertebral soft-tissue mass, short curve with severe apical rotation, intervertebral foraminal enlargement.[5] Other less common distinctive radiographic features, such as dural ectasia, spondylolisthesis, and defective pedicles. Familiarity with these deformities is, therefore, contributed to making an early diagnosis and optimizing treatment.[1]

CASE REPORT

A 15-year-old high school student who presented with 4 years history of upper back pain and hip pain, imbalance in her walking and 3 months history of some difficulty breathing on exertion. No trauma or falls and she is not a known asthmatic. Physical examination revealed multiple dark spots of varying sizes, about twelve on the trunk and upper limbs; with some nodules, about six on the upper body, S-shaped spine, prominence of the inferior angle and medial border of the right scapula, unevenly balanced hip region. Adams test was positive for this patient. Figure 1 shows the clinical pictures of the patient back with some features. X-ray revealed an s-shaped spine with Cobbs angle of 86 degrees in the thoracic spine and 56 degrees in the lumbar spine (Figure 2).

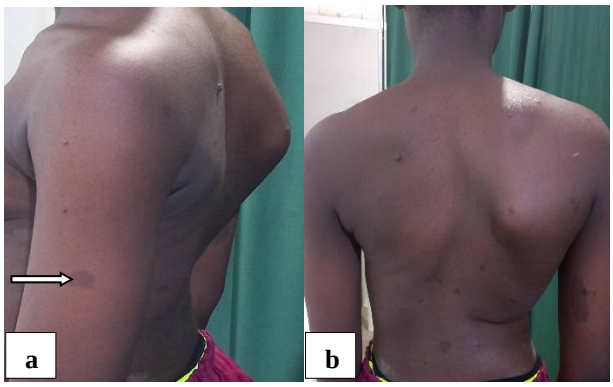

Figure 1: Clinical pictures of the back showing the lateral and anteroposterior views of the index patient showing (a) café au lait spot and (b) nodules.

Figure 2: X-ray of the whole spine anteroposterior and lateral views.

An assessment of adolescence idiopathic structural scoliosis coexisting with NF 1 was made. Patient and relatives were advised for a spine stabilization surgery to avoid worsening symptoms. Pre-operative bending X-rays, computed tomography (CT) and magnetic resonance imaging (MRI) of the whole spine were done to understand the anatomy of the spine and pedicles. Figure 3 shows the bending X-rays of the patient while Figure 4 shows the lumbosacral and pelvic X-rays indicating riser's sign (3/5). Patient was the reviewed by the team of anesthetists and was declared fit for surgery.

Intra-operatively, under general anesthesia and in prone position, incision and dissection was made to expose the T2 to L4 segments and then instrumentation of T3-L4 was done. Wake up test done and confirmed positive. Rods were then placed from distal to proximal segments. Intra operative blood loss was about 500 ml. She had one unit of blood transfused intraoperatively and another post operatively. Post-operative intensive care unit (ICU) care for 24 hours followed by post-operative check X-rays (Figure 5).

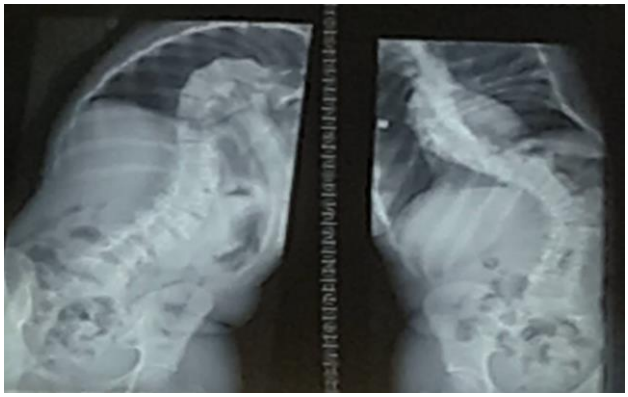

Figure 3: Bending X-rays of the index patient.

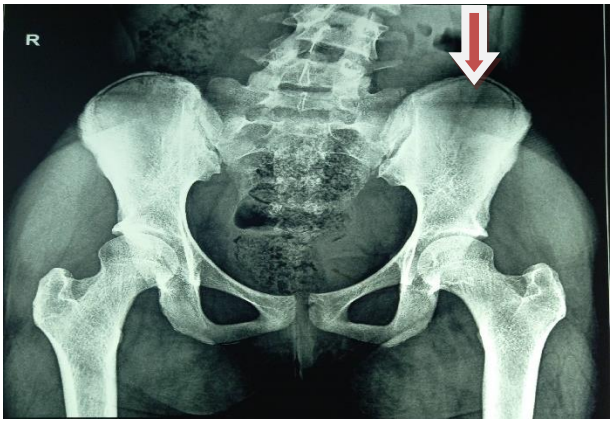

Figure 4: Lumbosacral and pelvic X-rays indicating riser's sign (3/5).

Thoracolumbar corset commenced and patient was made to ambulate about 24 hours post operatively with Zimmer's frame and was well tolerated. Alternate day wound dressing was done for the patient and after about 2 weeks

wound staples were removed and dressing discontinued. Figure 6 shows the post-operative clinical pictures with scars and reduced curvature of the patient's back. Patient was discharged from in-patient care about a week after surgery and then scheduled for follow up on out-patient basis.

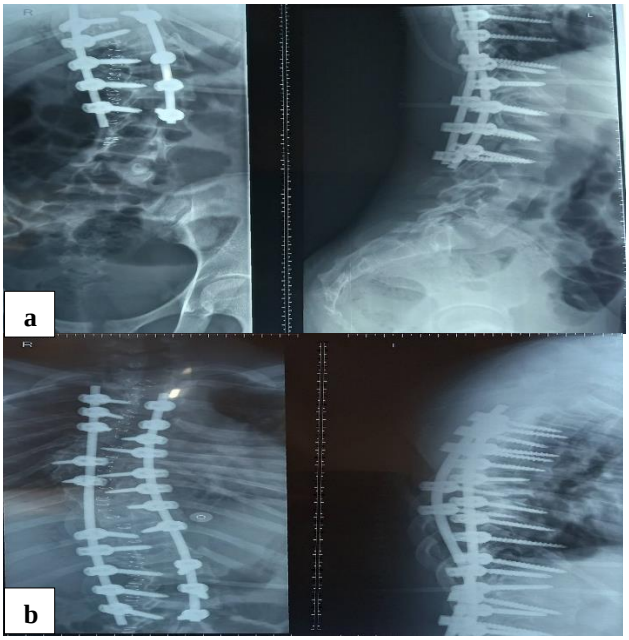

Figure 5: (a) Post-operative check X-rays; and (b) Post-operative check X-rays showing the whole extent of the implants (AP and lateral).

Figure 6: Post-operative clinical pictures showing anteroposterior and lateral views of the patient's back with surgical scar and reduced curvature.

DISCUSSION

Neurofibromatosis presenting with just skin lesions are poorly reported in this part of the world as skin lesions alone are not life threatening and do not affect quality of life significantly. In the index patient, the main reason behind presentation was the worsening spine curvature and the surfacing of some breathing difficulties. However, the skin lesions were not of primary concern to the patient and parents. NF 1 is a genetic condition that causes tumors to grow along the nerves. The tumors are usually benign but usually cause a range of symptoms. Neurofibromatosis could manifest with cutaneous and neurological features.[3] As earlier mentioned, scoliosis quite commonly coexists with NF 1 (as in the index patient as compared to other possible skeletal anomalies).[14]

NIH diagnostic criteria for NF 1

Clinical diagnosis based on presence of two of the following given in Table 1.

Table 1: National Institute of Health: diagnostic criteria for NF 1.

S. no.	Criteria
1	Six or more café-au-lait macules over 5 mm in diameter in prepubertal individuals and over 15 mm in greatest diameter in post pubertal individuals
2	Two or more neurofibromas of any type or one plexiform neurofibroma
3	Freckling in the axillary or inguinal regions
4	Two or more Lisch nodules (iris hamartomas)
5	Optic glioma
6	A distinctive osseous lesion such as sphenoid dysplasia or thinning of long bone cortex, with or without pseudarthrosis
7	First-degree relative (parent, sibling, or offspring) with NF-1 by the above criteria

To make a diagnosis of NF 1, two or more of the above-mentioned criteria must be met.[4] In the index patient, criteria 1 and 2 were present on examination thereby significantly confirming the diagnosis of NF 1 together with a Cobb's angle of more than ten degrees in both thoracic and lumbar regions.

Scoliosis is a term derived from the ancient Greek word "Skolios" which means curved or crooked. The term was first established by Galen between 130-201 AD.[7] Scoliosis as we know it now is a medical condition characterized by the chronic presence of substantial lateral curvature in a given region of the spine and is usually accompanied by rotation of the vertebrae within the curve.

The commonest type is the idiopathic scoliosis that often occurs in children from the age of eleven, hence the name idiopathic structural adolescent scoliosis. Due to the early growth spurt in girls during puberty, scoliosis is most commonly seen in adolescent girls.[7] Adolescent idiopathic scoliosis has a prevalence of 0.47-5.2% with the female to male ratio ranging between 1.5:1 and 3:1. These figures increase with increasing age.[7] Scoliosis can be divided according to etiology into idiopathic (adolescent or adult), congenital or neuromuscular.

The diagnosis of idiopathic scoliosis is made when all other causes are excluded and it is made up of 80% of all cases. Idiopathic scoliosis is further divided to adult and adolescent idiopathic scoliosis. Of important note is the fact that scoliosis could be of adult onset commonly in the face of degenerative spine diseases. Spinal deformity is the commonest orthopaedic manifestation in NF 1. Children with NF 1 develop one of two forms of scoliosis; dystrophic or non-dystrophic scoliosis.[13] Dystrophic scoliosis, on the other hand is a form of scoliosis that occurs due to bony changes related to neurofibromas affecting the spine. Dystrophic scoliosis is identified by picking up specific features on X-rays of the spine. It presents with abnormally thin ribs, weakened vertebral bones and severe spinal curvature including kyphosis and rotational deformities and is often associated deformities and is often associated with dural ectasia.[1,13] Non-dystrophic scoliosis, even in children with neurofibromatosis is quite similar to the adolescent idiopathic scoliosis.[1,13]

Cobb's angle

Dr John Cobb invented the Cobb's angle in 1948 as a standard of measurement for the determination and tracking of the LATERAL curvature of the spine (scoliosis).[8] Cobb suggested that to measure the angle, the upper border of the upper vertebrae; where the curvature seems to have begun; and the lower border of the lower vertebrae, where the curvature ends be taken as reference points in a plain radiograph. A parallel line to these reference points is drawn with another perpendicular line drawn across both lines to cross each other. The angle between these perpendicular lines is the angle of the curvature.[8,9] This method of measurement, though highly instrumental, has quite a number of limitations as it is highly subjective. Intra and inter-observer errors could affect the angles significantly.[8-10]

Wake up test

One of the most feared complications of spine surgery is paraplegia. The scoliosis research society in a case series that involved over 30,000 patients, the incidence of partial or complete paraplegia was 0.6%.[11] The Stagnara's wake-up-test, is a very easy way to detect voluntary motor function of the lower limb. No know false negative wake up test as at time of report.[12] Wake up test can be used alone or in combination with electrophysiological monitoring.[11,12] The use of a combined monitoring, intraoperative awakening and electrophysiological techniques would be optimal; Stagnara's test alone, however, performed with an anesthetic technique allowing a rapid recovery to a level of consciousness, is a reliable and practical method to detect as soon as possible neurological problems during major spine surgeries like scoliosis.[11,12] Contraindications to the test are mental retardation, psychological problems or preexisting neurological impairment and language.[14] Patient must have been primed preoperatively and informed or educated on the process.

In the reported case, the wake-up test of Stagnara was performed with the help of the anesthetist. Reversal of anesthesia was done to a reasonable level of consciousness where the patient could obey command and was asked to move her legs and hands and the test was positive.

Post-operative care and ambulation

Immediately following surgery, the index patient was transferred to the intensive care unit (high dependent unit) for proper supportive care and close monitoring for the first twenty-four to forty-eight hours. Graded oral intake was commenced the following day beginning with fluid-based diet and then gradually stepped up to full diet. Ambulating the patient was quite straightforward with Zimmer's frame and a thoracolumbar corset. Limitations to ambulation would have been poor pain control, complications arising from surgery (paraplegia, respiratory insufficiency), patient's psychology and many more which were all taken care of. Post-operative parenteral antibiotics and proper wound care we ensured in the patient to avoid wound breakdown or infections.

CONCLUSION

Scoliosis is the commonest manifestation of NF 1 with different severities and degrees of Cobb's angle. An assessment of scoliosis is made when the Cobb's angle greater than 10 degrees and Cobb's angles of less than 10 degrees will only need conservative management and will usually not need any surgical intervention. Adolescent females are mostly affected and management is dependent of degree of curvature, comorbidities like NF 1 and type of scoliosis.

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REFERENCES

1. Zhao C-M, Zhang W-J, Huang A-B, Chen Q, He Y-L, Wei Zhang, Yang H-L. Coexistence of Multiple rare spinal abnormalities in Type 1 Neurofibromatosis: A case report and Literature Review. Int J Clin Exp Med. 2015;8(10):17289-94.
2. Crawford AH, Bagamery N. Osseous manifestations of neurofibromatosis in childhood. J Pediatr Orthop. 1986;6:72-88.
3. Friedman JM. Epidermiology of Nuerofibromatosis Type 1. Am J Med Genet. 1999;89:1-6.
4. Neurofibromatosis Conference Statement. National Institute of Health consensus Development Conference. Arch Neurol. 1988;45:575-8.
5. Athanasios IT, Asif S, Hilali M. Spinal deformity in Neurofibromatosis type 1: diagnosis and treatment. Eur Spine J. 2005;14(5):427-39.

6. Durrani AA, Crawford AH, Chouhdry SN, Saifuddin A, Morley TR. Modulation of spinal deformities in patients with neurofibromatosis type 1. Spine (Phila Pa 1976). 2000;25:69-75.
7. Koineczny MR, Senyurt H, Krauspe R, Epidemiology of Adolescent Idiopathic Scoliosis. J Chils Orthop. 2013;7(1):3-9.
8. Musculoskeletal Consumer Review. Cobb angle and scoliosis. Available at: https://www.coreconcepts.com.sg/article/cobb-angle-and-scoliosis/. Accessed on 11 February 2022.
9. Tan J. Measuring the Cobb angle and scoliosis. Available at: http://www.health-articles.co.uk/measuring-the-cobb-angle-and-scoliosis/. Accessed on 11 February 2022.
10. SOSORT guideline committee, Weiss HR, Negrini S, Rigo M, Kotwicki T, Hawes MC, et al. Indications for conservative management of scoliosis. Scoliosis J. 2006;1:5.
11. Rodola F, D' Avolio S, Cherishing A, Vagnoni S, Forfe E. Wake up test during major spinal surgery under Remifentanil balanced anaesthesia. Eur Rev Med Pharm Sci. 2000;4:67-70.
12. Vauzelle C, Stagnara P, Jouviroux P. Functional monitoring of spinal cord activity during spinal surgery, 1973. Clin Ortgop Rel Res. 1973;93:173-8.
13. Akbarmia BA, Gabriel KR, Beckman E, Chalk D. Prevalence of scoliosis in Neurofibromatosis. Spine (phila Pa 1976). 1992;16:S244-8.
14. Riccardi VM. Neurofibromatosis: clinical heterogeneity. Curr Probl Cancer. 1982;7:1-34.

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