International Surgery Journal
Jain V et al. Int Surg J. 2017 Apr;4(4):1466-1468
http://www.ijsurgery.com
pISSN 2349-3305 | eISSN 2349-2902

DOI: http://dx.doi.org/10.18203/2349-2902.isj20171161

Case Report

Plexiform neurofibroma with AV malformation: amputation as treatment

Vikas Jain[1]*, Ved Prakash[2]

[1]Department of Oncosurgery, [2]Department of General Surgery, Asian Institute of Medical Sciences, Faridabad, Haryana, India

Received: 16 January 2017
Revised: 21 February 2017
Accepted: 23 February 2017

***Correspondence:**
Dr. Vikas Jain,
E-mail: drvickyjain@gmail.com

ABSTRACT

Neurofibromatosis with arteriovenous malformation is not very common and can results in complications, making it treatment more challenging and futile. Young man with complicated neurofibromatosis with limb deformity and joint destruction by arteriovenous malformation with life threatening syncopal attacks and generalized weakness is a rare presentation in our case. Multiple surgeries with recurrent symptoms and destructed limb makes the conservative surgery futile, hence amputation is considered best in such circumstances by authors. Amputation can be considered in symptomatic patient with gross limb deformity in such case for better quality of life.

Keywords: AV malformation, Amputations, Congenital, High output, Neurofibromatosis

INTRODUCTION

NF patients usually do not need any specific treatment except cases with significant complications. AV malformation is a known but rare complication in neurofibromatosis patient due to similar genetic deformity of tissue integrity.[1] These AV malformations may cause high output heart failure, hemorrhage as well as size and pressure related deformity in skeleton which need timely management before occurrence of fatal incidents. This case discusses management of such complication simultaneously.

CASE REPORT

40 years' male presented with rapidly progressing swelling for few days with sudden transient irresponsiveness of right lower limb, sudden dizziness and breathlessness. There were no complaints of difficulty in walking or any sensation loss previously.

Previously he was operated 16 years back for similar swellings 5 times, which was told benign last time but no documented details were available. Patient told that he had similar attack of breathlessness and syncope 3 years back also, for which he was managed symptomatically and started on ATT for suspected pulmonary tuberculosis. For small knee swelling on that time, he was suggested to wear clippers to support the knee joint, which he ignored and didn't compliant with advice.

On examination (Figure 1) huge 18 cm irregular swelling around the right knee joint without obvious joint line extending lower third of thigh to upper 1/3 of tibia region encircling the whole girth of lower limb with maximum diameter of around 10 cm. Swelling is irregular non-compressible, non-tender, soft to firm consistency, with raised local temperature. No bruit heard over swelling. There were prominent veins in skin over the swelling, but no ulceration. Vertical incisional scar present on medial side of swelling.

Similar small 5 cm hard bony swelling with incisional scar mark present distally on same limb. No regional lymphadenopathy.

Notable findings of multiple neurofibromas with café au lait spots seen on whole body. Similar neurofibromatosis lesion also seen in his brother too.

Figure 1: Clinical presentation of NF as multiple cafe AE' late spots with deformed knee joint.

Investigations

MRI knee joint lateral subluxation of distal femur over tibia with loss of congruity of lateral tibio-femoral joint space with bony deformity of distal femur with bowing of bones (Figure 2). Extensive secondary degenerative changes seen as florid osteophytes. Extensive soft tissue hypertrophy around knee joint and distal thigh. Large synovial joint effusion with synovial hypertrophy involving supra and pre-patellar compartment. Multiple abnormal vascular channels in defined areas.

Figure 2: MRI knee joint: destructed assembly.

Peripheral CT angiography

Peripheral hyper vascular soft tissue swelling in right mid, distal thigh, knee and leg region with multiple feeding vessels arising from distal SFA and other peripheral limb arteries. Suggestive of large AV malformation. Right femoral and tibial veins are dilated and tortuous. Distal shaft shows internal fixation plate in situ (Figure 3). Chest CT is suggestive of multiple infective nodes. No mass lesion. 2D echocardiography: Prolapse of anterior leaflet of tricuspid valve, otherwise normal.

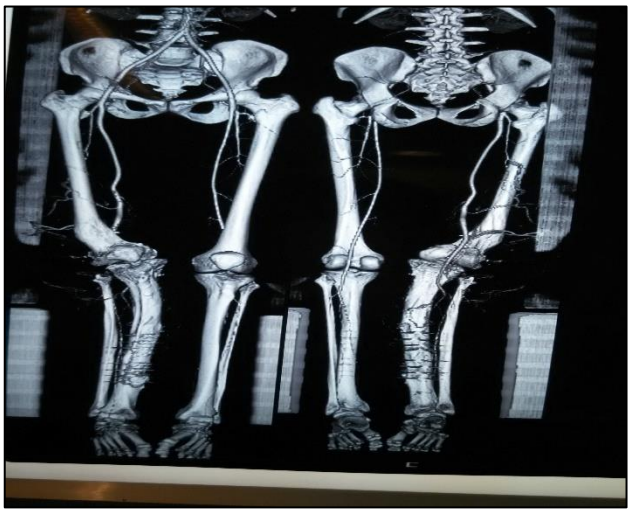

Figure 3: Peripheral CT angiogram: both lower limbs.

Finally, in view of history of recurrent syncopal attacks with valve prolapse, it was taken as semi emergent condition for high cardiac output status of AV malformation and amputation was considered for even NF like benign condition as life saving measure in already destroyed and degenerated lower limb.

DISCUSSION

Neurofibromas is a common genetic syndrome with multiple manifestations and its complication varying in different individuals. Plexiform neurofibromas are benign peripheral nerve sheath tumor affecting multiple trunk or fascicles of large nerve which is locally invasive and highly vascularized appearing in childhood, rarely in adolescence. Vascular complications are rare and again varies in its presentation. In one study of 31 patients of NF-1 with vascular abnormalities, 76 vascular abnormalities were identified and only 5 AV (6%) malformation were noticed and managed surgically.[1] Other asymptomatic abnormality were treated conservatively concluding that requirement of treatment depend on site of AVM and symptoms.

Most of these AV malformations have been reported in vertebral column, and central nervous system and rarely in scalp, intercostal space, peripheral extremities. These

sites are prone for severe hemorrhage and calamities.[1-4] Surgical treatment is considered in such cases associated with pain, neurological deficit, disfigurement, involvement of adjacent structures, and suspicion of malignancy which includes resection of tumors or correction of deformity.[5]

In our case, plexiform neurofibromas in extremity with recurrent lesions on same site and multiple surgery made the further curative resection extremely difficult with risk of heavy intra operative bleeding. Due to syncopal attacks and high cardiac output status and bony degeneration with deformity, conservative surgery or resection of tumor was deferred. Above knee amputation was considered with prosthesis for better quality of life. In any previous case reports, amputation and rehabilitation was never considered as surgical option which can provide better asymptomatic life and quality walking than before.

CONCLUSION

In grossly degenerated bones/joints by huge NF, resection of tumor alone cannot provide quality of life. Amputation should be considered along with rehabilitation by prosthesis for better prognosis and patients' comfort. Whenever surgical treatment is indicated, amputation should also be considered in treatment plan although not on priority.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

REFERENCES

1. Oderich GS, Sullivan TM, Bower TC, Gloviczki P, Miller DV. Vascular abnormalities in patients with neurofibromatosis syndrome type I: clinical spectrum management and results. J Vasc Surg. 2007;46:475-84.
2. Palazzuoli A, Pellegrini M, Ruocco P, Nuti R. A multidisciplinary approach in neurofibromatosis. Lancet Neurol. 2015;14(1):30-1.
3. Sidram V, Chandrakumar PC. Extracranial giant AVM of the scalp with plexiform neurofibromatosis in a juvenile child: a case report. Int J Health Sci Res. 2012;2(7):97-101.
4. Kim HJ, Seon HJ, Choi S. Ruptured aneurysm of Intercostal atriovenous malformation associated with neurofibromatosis type 1: a case report. Cardiovasc Intervent Radiol. 2011;34:74.
5. Dias IS, Pessoa SGP, Macedo JE, Cavalcante DJ. Surgical correction of a giant neurofibromas. Rev Bras Cir Plást. 2012;27(2):336-9.

Cite this article as: Jain V, Prakash V. Plexiform neurofibroma with AV malformation: amputation as treatment. Int Surg J 2017;4:1466-8.