Management of complicated neurofibromatosis our experience

Shahid Iftekhar Sadique[1*], Rajya laxmi Edupuganti[2], A.K. Chowohuri[3]

Dept. of Neurosurgery, Institute of Post Graduate Medical Education and Research, Kolkata, India

***Corresponding Author: Shahid Iftekhar Sadique**
Email: sisadique002@gmail.com

Abstract
Neuro fibromatosis is a autosomal dominant inherited disorder, with the classical 6 criteria for diagnosis it. many of these patients present with varied complications, we dealth with rare ones and preenting here.

Keywords: MMC, CSF, MRI.

Introduction
Neurofibromatios has been a well known genetic disorder from 18 th centruary. There has been extensive work on neurofibromatosis which lead to multiple criteria for its classification. NF are known for its complications varying from gross disfiguiring to multiple intra cranial and spinal space occupying lesions and early age of onset of saroma.

Materials and Methods
This is a prospective study done at BIN during the period 2016-2019.
Total no of patients with NF – 50
NF 1 -45
NF 2- 5
Complicated NF1 -3.

Case Discussion
Case 1
16 year old male with progressive swelling over left neck, pain over it for 3 months, progressive weakness of left half of body, with impairement of pain and temperature of left half of body. Father presented with multiple neurofibromatosis all over the body, sister had meningocoel repair at childhood. clinical diagnosis of brown; sequerad syndrome.

Mri Whole spine with brain screening: Large deep fascial tumour of left side of neck with C5-C6 neuroforaminal extension with cord compression.
OT: 1 st stage: Neck exploration with total removal of tumour.
2 nd stage: posterior cervical laminectomy with removal of tumour.
Post op: Transient quadriparesis followed by recovery.
BIOPSY: HPE +IHC - Malignant peripheral nerve sheath tumour
Malignant peripheral nerve sheath tumour are known entities, usually occurs in 40-50s age. In our case presence of NF is a predisposing factor.

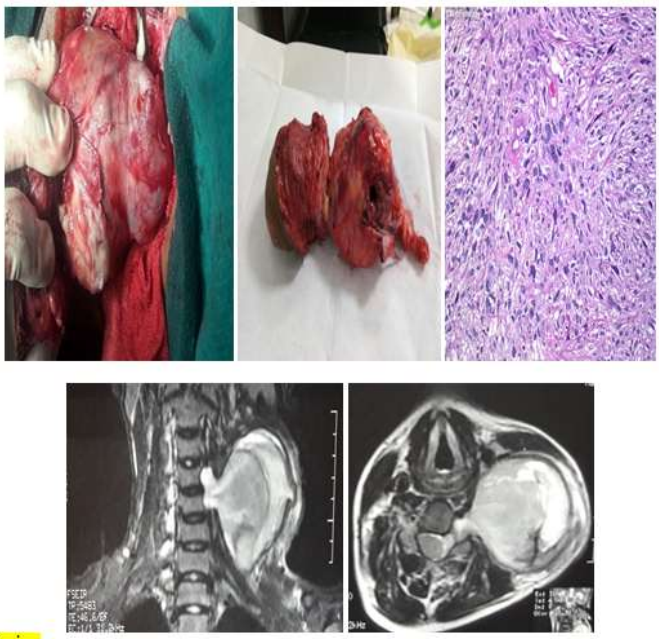

Fig. 1: operative specimen with slide with mri features

Case 2

35 year old military worker presents with progressive spastic quadriparesis over 4 months, father presented with multiple neurofibromatosis.

MRI whole spine with brain screening: multiple intra and extra spinal space occupying lesions present almost occupying all the vereteral levels with neuroforminal extension, maximal compressive affect seen at C4 to C6 level.

OT: posterior laminectomy with complete removal of sol done.
BIOPSY: Neurofibroma.

It is quite rare to find a scenario like above where almost whole of the vertebral levels are occupied with sols.

Fig. 2: multiple skin lesion demonstrated with MRI

Case 3

48 year old male labourer presents with slowly progressive swelling over back with progressive paraparesis, with positive NF 1.
Local examination shows a huge hard swelling over the back.
MRI whole spine with brain screening: large subcutaneous swelling with multiple dosal neuroforminal extention D 7-L2 with dorsal scoliosis.
OT: Stage 1: removal of subcutaneous swelling.
Stage 2: posterior dorsal laminectomy with total removal of sol with trans pedicular screw rod fixation.

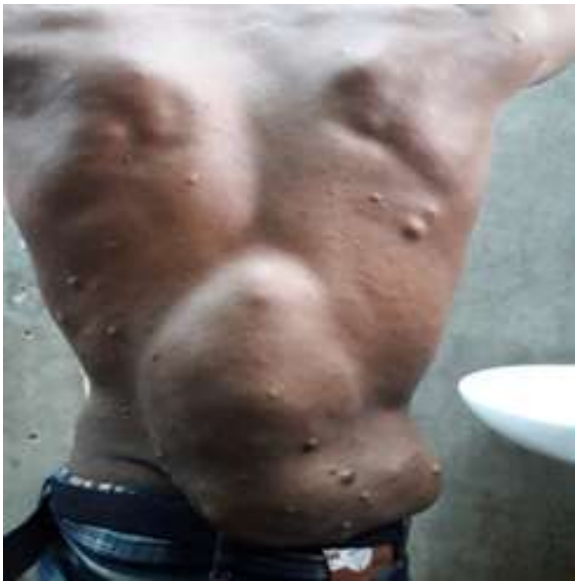

Fig. 3: large plexiform neurofibroma which turned in to sarcoma.

Discussions

Neurofibromatosis is a autosomal dominant inherited genetic disorder with varied clinical manifestations with variants in the form of incomplete to complete penetration. NF 1 is a tumour suppressor gene located on chromosome 17, mutation of this can manifest with wide variety of tumours ranging from skin, muscle, glial, meningeal, nerve sheath tumours.

Most of the patients harbor may silent growing tumours since childhood, manifesting with neurological deficits from a single tumour with largest size, compressive effect.

Decision making of neuro surgeon is crucial in the way that many patients still persist to have multiple tumours and resection of all these are not a possibility, thus surgical management is focused on tumours with maximal compressive affect with correlation of neurological symptoms.

Many of these features have an important age factor; for example, cutaneous neurofibromas do not usually develop before adolescence, and their frequency increases with age.

Neurological manifestations include involvement of the cerebrum, spine, cranial nerves and peripheral nerves.

Although the overall severity of NF1 is considered to increase with age and life expectancy is reduced in NF1 patients, the natural history of some potentially disabling or life-threatening NF1 complications remains unclear in adults.

The NF1-related neurological morbidity and prognosis in adult patients needs to be defined more precisely.

It is still unknown why few patients manifest with multiple spinal tumous only, mulipl meningioma only. still sub genetic pathways is underway.

Neurofibromas are futher classified as sub cutaneous, plexiform, diffuse, recurrent varieties.

In the above case series we described 3 cases of complicated neurofibromatosis.

CASE 1 have incompletely penetrant neurofibromatosis, with his only manifestation being cervical malignant peripheral nerve sheath tumour (neuro fibro sarcoma). He did not fit in to adult criteria of classical neurofibromatosis, but with positive family history. It shows the incomplete penetration of gene and the subtle manifestations of Nf with abrupt manifestation of MPNST.

CASE 2 Presents with isolated spinal neurofibromatosis. This patient presented with neurofibromas almost occupying all the the vertebral levels, but compressive affect only at a focal tumour.

We conducted a study over 2 year period, a prospective study, over 50 patients with neurofibromatosis, most of them fall in to classic / incomplete NF.

Here we presented with 3 cases of complicated neuro fibromatosis, their clinical presentation, our way of approach to the disease, and management of the complication of disease.

Plexiform neurofibroma on the face can also cause facial asymmetry.

The morbidity of plexiform neurofibromas in NF-1 is high, as they tend to grow up to a great size, producing disfigurement. Moreover, the risk of malignization is between 2 and 5%.

Multiorgan occurrence of NF1 requires a multidisciplinary approach. As there is no medical treatment for NF1, the management must be toward prevention and control of the complications. Although the rate of malignant transformation of NF1 is low (3 – 5%), these neoplasms can cause other clinical problems, including esthetic and functional compromising

The main peripheral nerve sheath tumors are characterized by neoplastic proliferations with Schwann cell differentiation. For instance, the Schwann cell represents the primary neoplastic cell component of neurofibroma characterized cytologically by wavy nuclear contours and S-100 protein expression.

Conclusion

Neurofibromatosis is a well known entity from many decades, with varied clinical manifestations. Due to the incomplete penetration of the gene varied manifestation which are typical of the lesion might be missing, but to present at an younger age with rare complications like above. Decision making by a neurosurgeon is very crucial as varied tumours might actually occur at a stance with a chance of developing new tumour still persist,with also high risk of developing malignancy in the same tumours too.

Conflict of Interest: None.

References

1. https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3491339/.
2. https://jmg.bmj.com/content/jmedgenet/39/5/311.full.pdf
3. https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3629555/
4. Agaram NP, Prakash S, Antonescu CR. Deep-seated plexiform schwannoma: a pathologic study of 16 cases and comparative analysis with the superficial variety. *Am J Surg Pathol* 2005;29:1042–1048.[PubMed] [Google Scholar]
5. Ducatman BS, Scheithauer BW. Malignant peripheral nerve sheath tumors with divergent differentiation. *Cancer* 1984;54:1049–57.
6. Zheng H, Chang L, Patel N, Yang J, Lowe L, Burns DK, Zhu Y. Induction of abnormal proliferation by nonmyelinating schwann cells triggers neurofibroma formation. *Cancer Cell* 2008;13:117–128.
7. Coffin CM, Cassity J, Viskochil D, Randall RL and Albritton K: Non-neurogenic sarcomas in four children and young adults with neurofibromatosis type 1. *Am J Med Gen* 2004;127A:40-43.

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