CASE REPORT

A RARE CASE OF NEUROFIBROMATOSIS TYPE 3

P. B. Sriram[1], A. ArunKumar[2], M. Natarajan[3], B. Palanikumar[4], K. Muralidharan[5]

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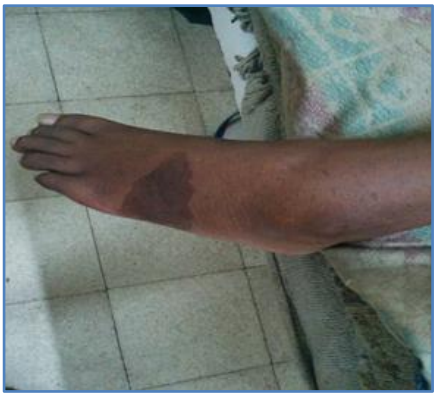
ABSTRACT: Neurofibromatosis is a type of genetic disorders characterized by cutaneous lesion and increased risk of brain tumours. NF1 is associated with more skin lesions and less CNS tumours, whereas NF2 is associated with minimal skin lesions and more brain tumours. This case is associated with both skin lesions and multiple CNS tumours.

KEYWORDS: Types of Neurofibromatosis café au lait spot. Freckling b/l vestibular schwannomas Riccardi classification.

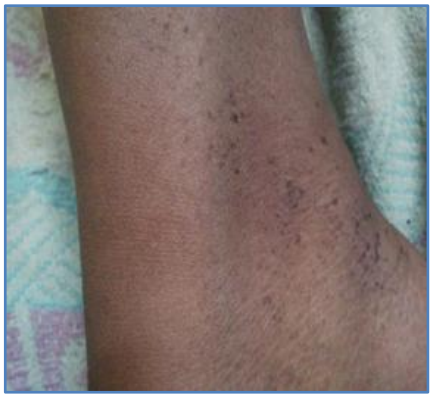
CASE SUMMARY: 17 years old female presented with complains of difficulty in walking for past 3years. She was apparently normal 3years back then she suddenly developed buckling of left knee while working in farm, after 2days she noticed difficulty in holding objects in left hand followed by involvement of right lower limb after 7days then followed by difficulty in using right hand after 2 days. During the same time period she gives history of bladder and bowel involvement in the form of involuntary micturition and defecation and history of sensory system involvement in the form of inability to feel cloth sensation, inability to feel hot and cold sensation and inability to feel pain was present. These symptoms remained static for 2years. After 2years there was apparent partial improvement of upper limb distal muscle weakness, sensory symptoms and bowel and bladder symptoms, history of minimal residual difficulty in mixing food or inability to hold objects persisted till now. History of inability to close left eye present since childhood and history of deviation of angle of mouth to right side present since childhood.

GENERAL EXAMINATION: Patient conscious, oriented, afebrile, not anemic, not icteric, no cyanosis, no clubbing, no pedal edema.

Skin lesions like lait spots, inguinal and plantar freckling, subcutaneous neurofibroma were present.



Lait Spot



Freckling

System examination:

MMSE score: 25/30

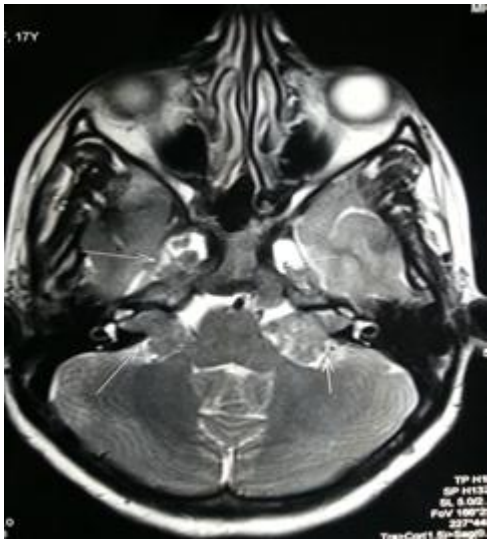
CN 1	N	N
CN 2	N	*Catarct +*
CN 3,4,6	N Pendular nystagmus+	*Abduction palsy +* *Pendular nystagmus+*
CN 5	N	N
CN 7		*LMN PALSY*
CN 8	RINNE - +ve WEBER – no lateralization	RINNE - +ve
CN 9,10	GAG +	GAG +
CN 11	N	N
CN 12	*TONGUE FIBRILLATION +*	*TONE & POWER NORMAL*

SUPERFICIAL REFLEX	RIGHT	LEFT
CORNEAL	PRESENT	PRESENT
CONJUCTIVAL	PRESENT	PRESENT
ABDOMINAL	PRESENT	PRESENT
PLANTAR	EXTENSOR	NO RESPONSE

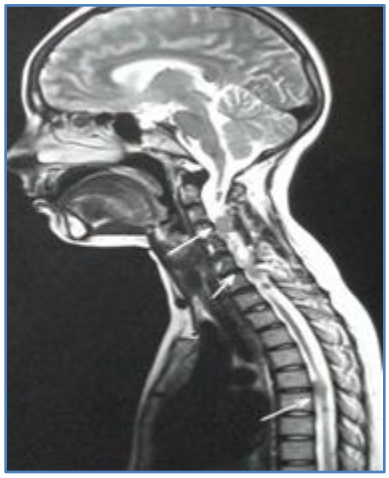
Tone: Normal in all limbs and reflexes are absent in all limbs. Examination of all other systems is normal.

Investigations: Blood sugar -70mg/dl, urea-27mgs%, creatinine-0.8mgs%. All other routine investigations were normal. ECHO-normal study, USG ABD-NAD.

MRI BRAIN SHOWED



b/l vestibular schwannomas



Multiple neurofibromas causing compressive myelopathy

Nerve conduction study: sensory motor axonal polyradiculopathy.
ENT: B/L minimal sensory neural hearing loss present
Eye: left eye showed posterior sub capsular cataract.

FUNDUS EXAMINATION:

1. Fibrous band extending from optic disc to macula.
2. Persistent hyperplastic primary vitreous extending from optic disc.

CONCLUSION: It's a case of Neurofibromatosis type 3 where it is associated with features of NF2 with skin and peripheral lesions.

RICCARDI CLASSIFICATION OF NEUROFIBROMATOSIS[1]:

- NF 1 - von recklinghausen's disease.
- NF 2 – Acoustic.
- NF 3 – mixed (multiple brain and spinal tumours with CALM and neurofibroma).
- NF 4 – variant (both neurofibroma and CALM are present but further Categorization is not possible).
- NF 5 – segmental (CALM and neurofibroma is limited to a Unilateral segment).
- NF 6 - familial CALM (multiple CALM Without neurofibroma).
- NF 7 - late onset (manifestation occur after 20 years, CALMs absent).
- NF 8 - not otherwise specified.

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AUTHORS:

1. P. B. Sriram
2. A. ArunKumar
3. M. Natarajan
4. B. Palanikumar
5. K. Muralidharan

PARTICULARS OF CONTRIBUTORS:

1. Post Graduate, Department of General Medicine, Madurai Medical College.
2. Post Graduate, Department of General Medicine, Madurai Medical College.
3. Professor, Department of General Medicine, Madurai Medical College.
4. Assistant Professor, Department of General Medicine, Madurai Medical College.
5. Assistant Professor, Department of General Medicine, Madurai Medical College.

NAME ADDRESS EMAIL ID OF THE CORRESPONDING AUTHOR:

Dr. P. B. Sriram,
No. 23, Bharathiar Salai,
Thilagar Nagar,
KaraiKudi-630002.
E-mail: srirambp@gmail.com

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