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Case Report

NEUROFIBROMATOSIS 1: A RARE PRESENTATION

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Abstract
A14 month's old female child brought to emergency department with the h/o abnormal behavior, not responding to parents & right UL & LL seizures since 1hr 40min. In view of ongoing seizures & intermittent desaturation, child was intubated & treated with multiple antiepileptic drugs as child was resistant to primary treatment. Child has multiple café au lait spots/macules of variable size& abnormal fat pads. Right LL thinner than left LL. Cutaneous findings favours diagnosis towards NF1[1]. MRI brain showed right cerebellar hypoplasia& MRI spectroscopy showed thalamic involvement[7]. Cerebellar hypoplasia is very rare in a case with NF1[3]. NF1 associated with cerebellar hypoplasia presenting with seizures is 55.5%[4]. Frequently in NF1, the presence of multiple CALMs is the first feature noticed in a child & other features do not appear later childhood, delaying the diagnosis[10]. Early airway management in patients of NF1 presenting with seizures is lifesaving. Early detection and prompt attention to complications may reduce overall morbidity and mortality [9].
Keywords: Seizures/Status epilepticus, Early airway management, Multiple antiepileptic drugs, Café-au-lait spots/macules, Cerebellar hypoplasia, Thalamic involvement, Early detection & prompt attention.

Introduction

NF1 or Von Recklinghausen disease is a multisystem neurocutaneous disorder and the most common phakomatosis[1], characterized by optical tumors and other central nervous system tumors, certain bony abnormalities, some learning deficits and increased risk of certain non-nervous system cancers. NF1 is usually noticed during childhood with the development of skin findings. But presence of cerebellar hypoplasia and seizures are rare with NF1. NF1 gene localized on chromosome 17q11.2, Autosomal dominant inheritance, extremely variable expression

Case Report

On 26/09/2014 at 12:40 pm a 14 months old female child brought to emergency

department with the h/o seizures in right UL and LL. It started with the H/o abnormal behavior since 11:00 am (26/09/2014), not responding to parents, right side focal convulsions [started with twitching of right eyelid, right side of face, right UL and then right LL]. On arrival to emergency at 12:40 pm, baby had complex right focal seizures [right UL & LL] with unconsciousness.
Inj LORAZEPAM 0.1mg/kgiv given
AIRWAY: patent, SPO2-75-85%, child was intubated in view of low conscious level, status epilepticus and intermittent desaturation and put on ambu ventilation
BREATHING: B/L chest rise present, b/l air entry equal, Ventilation rate of 30-35cpm
CIRCULATION: All peripheries warm & All peripheral pulses felt, CRT <2 sec, HR-200bpm, BP- 90/50 mm Hg, GRBS- 117 mg/dl, temp- 99.2 F, iv line secured
DISABILITY: GCS- ongoing seizures, pupils 2mm BERL, no external injuries noted,
DRUGS: Rectal diazepam given at nearby hospital at 12:25pm

Baby was still on seizure after giving

12:40pm- Inj Lorazepam 0.1 mg/kg

↓

Seizure persists

↓

12:50pm-Inj Fosphenytoin270mg i.v. over 20min

Seizure persists

↓

1:20pm- Inj sodium valproate 90mg over 20min

Seizure persists

↓

1:40pm- Inj Phenobarbitone 20mg/kg over 20min

Seizure persists

↓

2:10pm- Inj Levetiracetam 100mg over 20min

Seizure persists

↓

Started on Midazolam infusion 5micro gm/kg/min

↓

Seizures terminated at 2:45pm

Connected to ventilator SIMV-PC mode

Past medical/ surgical history: not on any medications
Birth history: uneventful

Immunization history: up to date
Developmental history: as per age
Family history: Nothing significant

On Examination:

Multiple hairy pigmented nevus on scalp face chest abdomen and extremities coalescing as a patch on lower abdomen and legs including the soles. Multiple café au lait macules on trunk and limbs. Hair, nails, mucosa normal. Abnormal fat pads on right hip & calf and b/l forearms

DTRs symmetrical, right knee difficult to elicit. Right LL thinner than Left LL

Right paucity of movement UL>LLTone-hypotonic

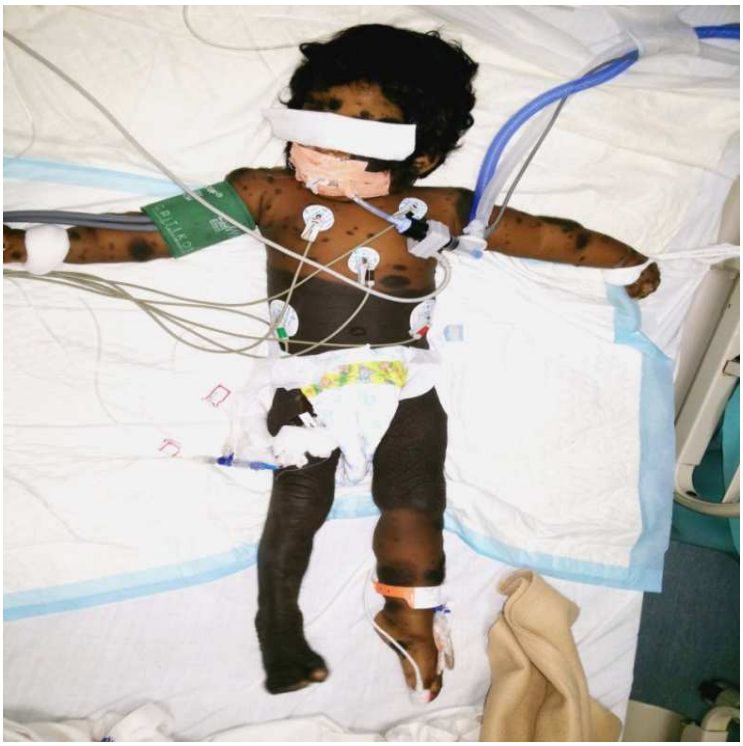
Investigations

- CBC: Hb 9.9g/dl, TC 10,250 platelet count 5,71,000 PCV 30%
- CRP: 48 mg/dl
- Serum electrolytes: Na 138/ K 4.5/ Cl 100 with Ca2+5.22 , Mg2+ 2.0, Phosphorus 7.4
- Serum Prolactin: 11.3ng/ml
- NH_3: 29microgm/dl
- Lactate: 0.9 mmol/lt
- ALP: 206 U/L
- Serum Creatinine: 0.3mg/dl
- USG abdomen: No significant abnormality detected
- MRI brain

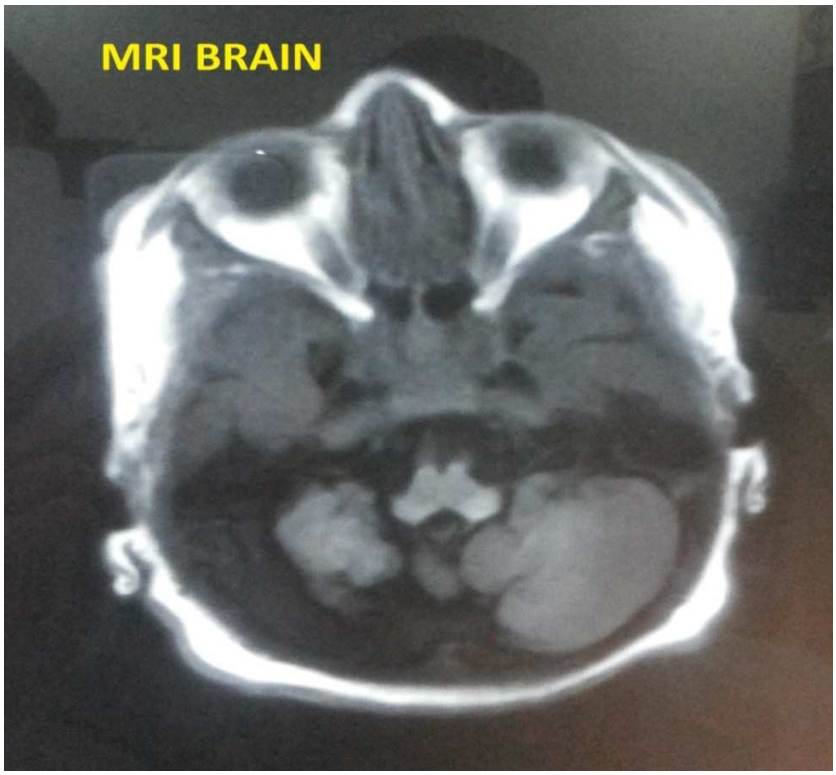
RIGHT CEREBELLAR HYPOPLASIA

- MR spectroscopy (right side)

DECREASED NAA/CHOLINE RATIO



Multiple hairy pigmented nevus on scalp face chest abdomen and extremities coalescing as a patch on lower abdomen and legs including the soles. Multiple café au lait macules on trunk and limbs. Right LL thinner than Left LL. Abnormal fat pads on right hip & calf and b/l forearm



Right cerebellar Hypoplasia

MR Spectroscopy (right)-Decreased NAA/ Choline ratio

Discussion

Seizures usually occur in 6-7% of NF1 individuals[2] and are usually resistant to primary antiepileptic treatment. Early airway management, maintenance of vitals & need of multiple antiepileptic drugs in the prime management of patients with NF1 presenting to emergency department with seizures. Cerebellar hypoplasia is very rare in a case with NF1[3]. NF1 associated with cerebellar hypoplasia presenting with seizures is 55.5%[4]. Moreover tendency toward abnormal EEG findings, associated in a minor percentage of cases with epilepsy is also evident[5]

Cerebellar hypoplasia should be considered in the young child presenting with developmental delay with prominent motor involvement, together with cerebellar signs &/or microcephaly[6]MRI is preferred for diagnostic head imaging. Hyper-intense lesions on T2-weighted brain MRI are probably caused by aberrant myelination or gliosis and are pathognomonic of NF1.Based on the neuropathological study by Dipaolo and colleagues, choline elevations reflect increased myelin turnover in areas of intramyelinic edema, which is followed by neuronal injury (reduced N-Acetyl Aspartate) [7]

The risk of an affected individual with NF1 transmitting the disease to their child is 50% but this cannot predict the severity of any inherited disease. Where parents have had the first affected child known in a family, both parents should be examined for cutaneous stigmata or Lisch nodules. Prenatal testing is possible using fatal DNA extracted from chronic villous sampling or from amniocentesis. Pre-implantation genetic diagnosis is also available. Genetic counselling prior to conception should be advised in all individuals with NF[8]. Most people with NF1 lead relatively long and healthy lives, but it does reduce life expectancy. Early detection and prompt attention to complications may reduce overall morbidity and mortality [9]

Conclusion:

Early airway management in patients of NF1 presenting with seizures is lifesaving. Early recognition of NF1 and followed with genetic counseling of the couples is of paramount importance in dealing with this disease.

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