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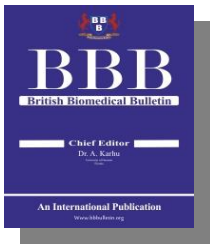

Case Report

Neurofibromatosis Type 1 with Chiari Malformation Type 1 in Child: A Case Report

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A R T I C L E I N F O

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A B S T R A C T

Objective: Importance of neuro imaging in Neurofibromatosis type I before doing lumbar puncture as Neurofibromatosis type 1 with Arnold Chiari malformation have been rarely reported in children.
Methods: 11 years old boy having neurofibromatosis type I presented with fever and convulsions. He was diagnosed treated for encephalitis Neuro imaging was done due to persistent symptoms.
Results: Neuro imaging detected Arnold chiari malformation type 1. Eventually, patient improved with treatment.
Conclusion: Neuro imaging should be done in all cases of Neurofibromatosis type I before doing lumbar puncture to prevent brain stem compression.

Introduction

Neurofibromatosis type1 (NF1) is an autosomal dominant neurocutaneous syndrome due to a mutation in chromosome 17q11.2. Its characteristic features include multiple cafe-au-lait spots, axillary and inguinal freckling, multiple cutaneous neurofibromas, and iris Lisch nodules.[1] De novo mutations occur in nearly fifty percent of cases. Association of NF1 with various cerebral dysplasias has been described.[2] Chiari malformation is one such unusual association. Arnold Chiari type I malformation (ACM1) is characterized by the displacement of the cerebellar tonsils into the upper part of the cervical canal. Association of NF1 and ACM1 has been rarely reported in paediatric age group.[3] We report a boy in whom ACM1 was detected during neuroimaging who had otherwise neurofibromatosis and encephalitis.

Case Report

An 11-year-old boy, second born to a non consanguineous parentage presented with fever, headache and vomiting, altered sensorium of one-day duration and one episode of generalised tonic clonic convulsion. Family history revealed neurofibromatosis in great grandmother, grandfather (Figure 1B), mother and maternal uncle. On examination, vitals were stable and his Glasgow coma scale was 13/15. He had multiple cafe-au-lait spots (figure 1A), iris Lisch nodules (Figures 2B).There was terminal neck stiffness. There was no evidence of papilledema. Other systemic examination was unrewarding. Blood counts, serum electrolytes, random blood sugar were within normal limits. Cerebrospinal fluid (CSF) analysis revealed 28 lymphocytes/ mm^3, protein 46mg/dl, sugar 56mg/dl, negative for gram staining and no bacterial and fungal growth after 48 hours of CSF culture. He was treated for viral meningo encephalitis and convulsions using 20% mannitol 5ml/kg 6^{th} hourly and phenytoin 20mg/kg loading dose followed by 8mg/kg in two divided doses .As patient continued to have persistent early morning headache with vomiting, magnetic resonance imaging (MRI) of brain and spine was done that showed tonsillar herniation with loss of normal cervical spine lordosis suggestive of ACM1 (Figure 2A). Electro encephalogram was normal. Symptoms subsided after 10 days of treatment with no neurological abnormalities. Auditory evaluation was normal and his IQ was found out to be 71. Parents were counselled regarding disease and advised yearly follow up for ophthalmological, spine, cardiovascular and neurogical evaluation and any new onset symptoms.

Discussion

NF-1 is more diagnosed now than in the past because of the use of rigid clinical criteria. The present case satisfied the diagnostic criteria for NF-1, having more than 6 cafe-au-lait spots measuring greater than 15 mm in diameter, Lisch nodules in the iris and presence of neurofibromatosis in the family members.

NF1 has various clinical manifestations. Central nervous system (CNS) lesions include optic gliomas, non-neoplastic hamartomas, plexiform neuro-fibromas, intraspinal neurofibromas and dural ectasia, macrocephaly, unilateral sphenoid dysplasia, various heterotopias, excessive myelination in gray matter, atypical glial cell nests, subependymal glial nodules, ependymal ectopias, intramedullary schwannosis, stenoses of the cerebral aqueduct, and hyper intense lesions (T2-weighted MRI) in the basal ganglia, internal capsule, and cerebellum.[3] Neurological manifestations are found in 40% of patients with neurofibromatosis.[4] However, the

association of NF-1 and ACM1 is very uncommonly reported.[3-5] ACM1 is either asymptomatic or symptomatic due to occipitalization of the atlas, scoliosis, spinabifida or syringomyelia. Only herniation of the cerebellar tonsils and loss of normal cervical lordosis was present in our patient.

The prevalence of ACM 1 is of 1 in 3700 and that of NF-1 is 1 in 4500 to 1 in 6700, and the probability of having both conditions is therefore very low (1:16 650 000 to 1:24 790 000).[3] ACM1 can be asymptomatic and can be detected as an incidental finding in neuro imaging studies, as described by Dooley J *et al*.[5] Hence the routine use of cranial-cervical MRI in any NF-1, irrespective of neurological symptoms and signs, will probably reveal a higher frequency of this association. Tubbs *et al* reported NF1 in 5.4% of 130 surgically addressed ACMs and ACM1 in 8.6% of 198 NF1 patients.[6]

Dysplasia and proliferation of ectodermal and endodermal tissue in NF1 is caused by gene mutation (17q11.2) where as pathogenesis of ACM1 is related to hypoplasia of the posterior fossa, leading to herniation of the cerebellum through the foramen magnum.[7] The abnormal embryonic development secondary to NF1 could be responsible for the posterior fossa hypoplasia found in ACM1.[8]

Neurological symptoms have been reported with most common symptom being Recurrent periodical headache.[3,5,9,10] Feature such as hemiparalysis[9], hydrocephalus[9], gait disturbance[10], dizziness[10], seizure[5], and learning disability[5] have also been documented. In our case, the boy had headache that later subsided on conservative management. Annual follow up for ophthalmological, spine, cardiovascular and neurological evaluation and after new onset symptoms is recommended.[1] Persistent or recurrent headache, pain, neurological deficits, syringomyelia necessitates posterior craniovertebral decompression.

Conclusion

Magnetic resonance imaging should be done before lumbar puncture in all cases of Neurofibromatosis type 1 irrespective of signs of raised intracranial tension and in presence of persistent or recurrent headaches.

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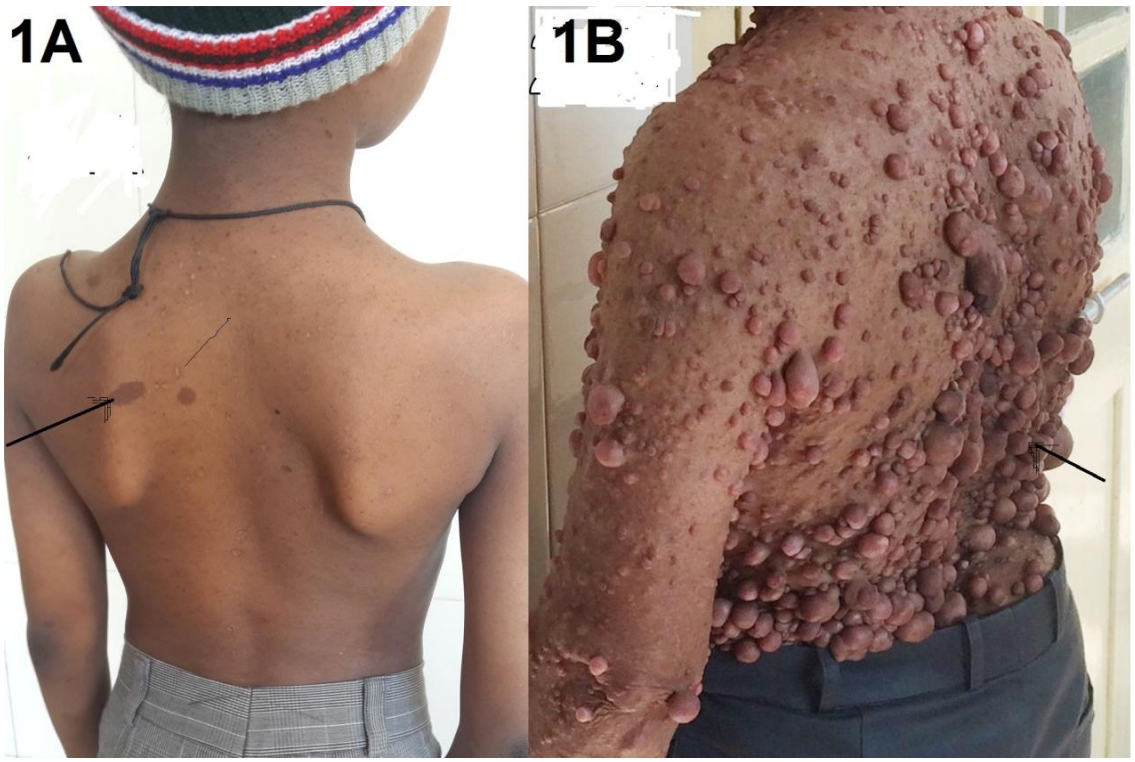

Figure 1. (A) Multiple cafe au lait spots in patient (arrow), (B) Neurofibromas in grand father (arrow)

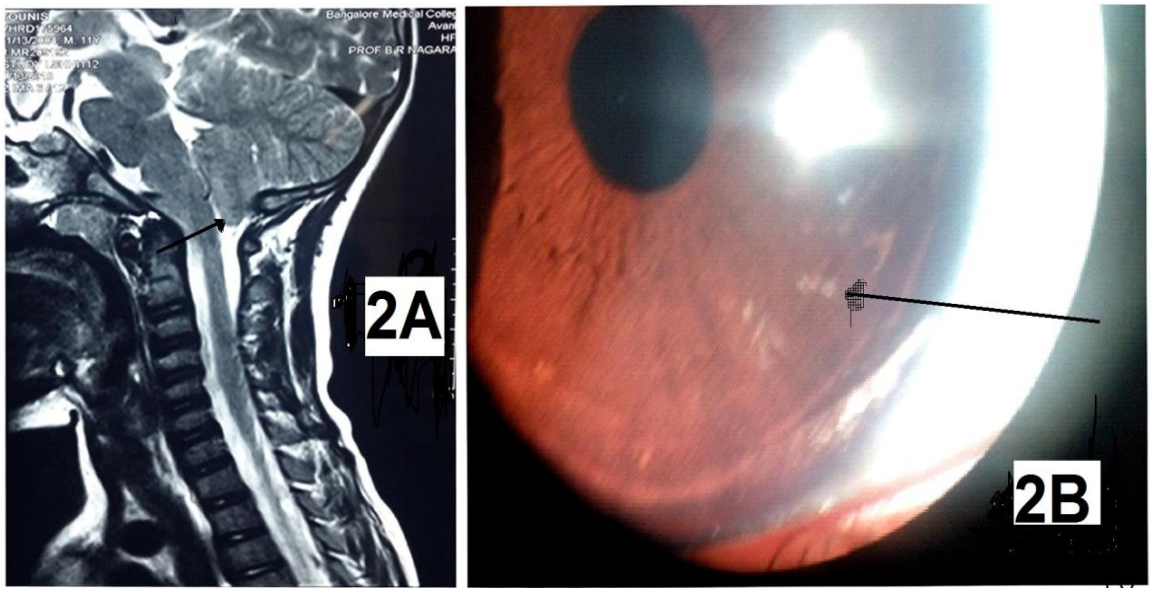

Figure 2. (A) MRI showing tonsillar herniation (arrow), (B) Lisch nodules (arrow)