Case Report
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CASE REPORTS

UNUSUAL LARGE BRAIN CYST IN NEUROFIBROMATOSIS

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ABSTRACT

Cystic lesions of brain have already been described in certain case reports as a rare finding in neurofibromatosis. CT scan sometimes shows large cysts with ring enhancement causing compression of the adjacent structures and simultaneously the patient is symptomatic.

We report a 10-year-old boy with neurofibromatosis type 1 demonstrating a single large cystic lesion in right parietal lobe of the brain with ring enhancement on CT scan with contrast. Histopathological evidence revealed the nature of the cyst as grade 3, anaplastic oligodendroglioma, which is an extremely rare manifestation of this multi systemic disorder. Along with multiple brown spots on his abdomen, the boy had frequent complaints of persistent headache and projectile vomiting on which this workup was done. Interestingly, one of his siblings was found to have similar cutaneous manifestations with absence of any neurological findings and normal CT scan.

Keywords: Café-au-lait spots, Neurofibromatosis, Neurocutaneous, Oligodendroglioma.

INTRODUCTION

Neurofibromatosis type 1 (NF1) is an inherited disorder characterized by formation of tumors (neurofibromas) involving nerve tissues in skin, subcutaneous tissues, cranial nerves, and spinal root nerves. It is relatively a rare pediatric disorder.

CASE REPORT

A 10 years old boy presented with complaints of persistent headache and vomiting for five days. Headache was severe and was associated with sweating. His vomiting which was projectile, early in the morning with no other associations had a frequency of seven episodes a day and measured approximately half a liter having yellowish tinge containing ingested food. There was no history of seizures or any focal neurological deficits.

His general physical examination revealed synorphysis, microcephaly, multiple café- au-lit spots (Fig.2) varying from 5 cm to 15 cm in size with certain hypo pigmented areas, axillary as well as inguinal freckling and certain ash grey macules in lumbosacral area and left thigh which led to his clinical diagnosis of Neurofibromatosis[1]. Surprisingly examination of eye showed normal iris and clear fundus. Cognition was subnormal than expected for age. CT scan brain with contrast revealed a large cystic lesion in right parietal lobe compressing thelateral sinus, however there was no midline shift (fig-1) and his electroencephalogram was also normal. Examination of the sibling also revealed café-au-lit spots and axillary freckling.

The boy underwent burr hole procedure and biopsy was taken from right parietal lobe by a neurosurgeon and was sent for histopathology which revealed "Anaplastic Oligodendroglioma grade 3. MRI was not advised due to obvious findings and confirmation of diagnosis on histopathology.

The boy was subjected to total removal of the tumor and treated with chemotherapy and radiotherapy using conventional strategy. His parents were thoroughly counseled about the prognosis, morbidity and mortality of the disease. During the last six months follow up,

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there is gradual paresis of left half of body probably due to necrotic changes in the tumor and compression effects. He is being managed by neuro-physician and neurosurgeon for paresis.

DISCUSSION

Most tumors associated with NF1 are benign (non-cancerous) skin tumors, which grow on nerves throughout the body. Neurofibromas (NFs) are the most common type of tumor in people with NF1, dermal (sub-cutaneous) neurofibromas and plexiform neurofibromas being the two major subtypes. Brain tumors are diagnosed in approximately 15%–20% of children with NF[1,2]. Astrocytoma is the major type of CNS tumors in NF1, and pilocytic astrocytoma (WHO grade I) the main histological subtype[3,4]. Although other rare types of CNS tumors have been reported in NF1, including ependymomas[5], me-dulloblastomas[6] and dysplastic neuroepithelial tumors[7], oligodendrogiloma is one of these rare tumors suggesting that the incidence of these tumors in NF1 is very low.

CT scan finding of large cystic lesion may be of variable etiology like brain abscess, hydatid cyst, brain infarction, and tumors with central necrosis like oligodendrogliomas, tuberculomas, and many more. Detailed evaluation by history, clinical examination and required investigations can give a clue to the exact diagnosis.

Neurofibromatosis type 1 has frequency of 1 in 4,000[8]. It is an autosomal dominant disorder consisting of nerve tissue tumors(i.e. neurofibromatosis) with mutation of neurofibromin on chromosome 17q11.2[9,10]. Neurofibromin (a GTPase activating enzyme GAP) is a tumor suppressor and functions to inhibit the p21 rasoncoprotein[8]. In the absence of the inhibitory control of neurofibromin on rasonco-protein, cellular proliferation is erratic and uncontrolled, resulting in unbalanced cellular proliferation and tumor development. The disorder affects all neural crest cells including schwann cells, melanocytes and endoneural fibroblasts. Cellular elements from these cell types proliferate excessively throughout the body forming tumors. Melanocyte proliferation result in disordered skin pigmentation and "café-au-lait" spots. The tumors may cause swelling beneath the skin, skeletal involvement, pressure effects on spinal nerve roots, and other neurological problems[9]. It affects males and females equally.

The diagnosis of NF1 is made if any two of the following seven criteria are met:

Two or more neurofibromas on or under the skin, or one plexiformneurofibroma (a large cluster of tumors involving multiple nerves); neurofibromas are the subcutaneous swellings characteristic of the disease, and increase in number with age.

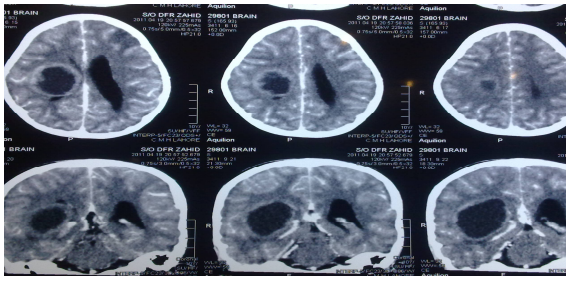

Figure-1: Large brain cyst with ring enhancement involving right fronto-parietal lobe compressing right lateral ventricle which turned out to be oligodendroglioma.

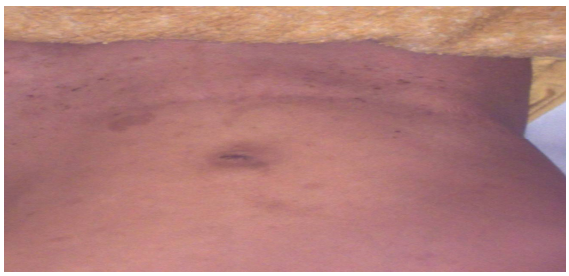

Figure-2: Multiple café-au-lit –spots on abdomen and upper.

Freckling of the groin or the axilla.

Café-au-lait spots, pigmented, light brown macules located on nerves, with smooth edges (coast of California) (12) http://en. wikipedia. org/wiki/Neurofibromatosis- cite_ note-rubin-0 birthmarks. Six or more measuring 5 mm in greatest diameter in prepubertal individuals and over 15 mm in greatest diameter in postpubertal individuals.

Skeletal abnormalities, such as sphenoid dysplasia or thinning of the cortex of the long

bones of the body (i.e. bones of the leg, potentially resulting in bowing of the legs)[8].

Lisch nodules (hamartomas of iris), freckling in the iris

Tumors on the optic nerve, also known as an optic glioma

Macrocephaly in 30-50% of the pediatric population without any hydrocephalus

Epilepsy (seizures)

Juvenile posterior lenticular opacity[9].

Sometimes tumor surgery is mandatory for the patient. Affected individuals may need multiple surgeries (reduction or Gamma knife surgery), depending on the location of tumor.

NF1, is a good example of the phenomenon of variable expressivity; differing severities of disease in different individuals with the same genotype. Both NF-1 and NF-2 can also appear to be spontaneous mutations, with no family history. These cases account for about one half of neurofibromatosis.

CONFLICT OF INTEREST

This study has no conflict of interest to declare by any author.

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