CASE REPORT

UNILATERAL OPEN LIP SCHIZENCEPHALY – A RARE CASE REPORT

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INTRODUCTION: Schizencephaly is a rare neuronal migration disorder with an incidence of 1.5 in 1,00,000 live births. Herein we report a rare case of left sided open lip schizencephaly.

KEYWORDS: neuronal migration, developmental delay, schizencephaly.

CASE REPORT: A 9 year old male child came to our OPD with complaints of right upper and lower limb weakness which was recognised by his parents at 6 months of age .he was a product of 2nd degree consanguineous marriage delivered through NVD which was a prolonged labour requiring forceps assistance, antenatal h/o was uneventful. He had a height of 118cm (0-1SD) and weight of 25kg (0-1SD). There was a mild developmental delay of gross motor domain, the child attained walking without support at the age of one and a half year and the weakness was non progressive. Child had 1 episode of unprovoked right sided focal seizure at 3 years of age. On examination he had uniform wasting of right sided upper and lower limb muscles with right sided facial nerve palsy of upper motor neuron type, power was 4/5 in both upper and lower limbs, he had slurring of speech, and his gait was circumductory. There were no involuntary movements. There was no mental retardation. Skull and spine were normal. There were no neurocutaneous markers. Keeping the above findings in mind work up for infantile hemiplegia was done, laboratory investigations revealed hemogram, serum electrolytes, ammonia and lactate which were in normal ranges. In order to rule out structural abnormalities MRI brain was done which showed a uniform grey matter lined cleft extending from pial surface of the cerebral mantle upto the ventricular ependyma.

Thus the diagnosis was clinched with the help of MRI scan. Prognosis was explained to the parents and physiotherapy was advised.

Close differentials include: other disorders of neuronal migration like focal cortical dysplasias, grey matter heterotopia, porencephaly etc.

DISCUSSION: Schizencephaly is a rare cortical malformation that manifests as a grey matter lined cleft extending from the pial surface of the cerebral mantle upto ventricular ependyma. Incidence is estimated to be around 1.5 in 1,00,000 live births. It is almost always sporadic and there is no known gender predilection. Developmental delay, motor disturbances correlate with degree of anatomical abnormality. Seizures are relatively common. Exact pathogenesis is uncertain but is most likely to be a disorder of neuronal migration. Some familial cases have been reported where heterozygous germline mutations of homeobox gene EMX2 are often encountered. It can be divided into two morphological types: 1. open lip, 2.closed lip. Most frequently the cleft involves posterior frontal and parietal lobes (70%). Large clefts can involve temporal and occipital lobes, isolated involvement is uncommon. MRI is the imaging modality of choice as it enables better differentiation of grey and white matter defects.

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T1 weighted image of MRI

T2 weighted image showing hyperintense uniform cleft extending from cerebral mantle upto the ventricular endyma.

Picture of the patient showing wasting of right sided upper and lower limb muscles.

Picture showing wasting of muscles of the back.

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