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A Case Of Hemimegalencephaly With Hypomelanosis Of Ito

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Abstract : Hypomelanosis of Ito (Incontinentia pigmenti achromicans) is a rare phakomatosis characterised by hypopigmented patches or whorls along the (lines of Blaschko) with involvement of central nervous system,eyes and musculoskeletal structures. Very few reports of its association with hemimegalencephaly are reported. Reporting a 31 day female baby born for non consanguineous parents presented with refractory seizures. The child had a hypopigmented patch over left temporal region ,was clinically diagnosed as Hypomelanosis of Ito. MRI brain revealed the malformation. The case is reported for its rarity and its rarer association.

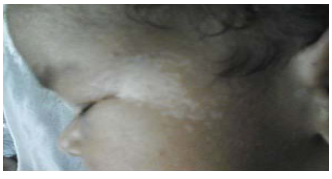
Keyword : Hypomelanosis Of Ito, Hemimegalencephaly, Refractory seizures.

INTRODUCTION

Hypomelanosis of Ito/Incontinentia pigmenti achromicans is a rare phakomatosis characterised by hypopigmented patches or whorls along (lines of Blaschko) with involvement of central nervous system, eyes and musculoskeletal structures. .Frequency is about 1in 80000 to 1 in 100000. The association with hemimegalencephaly is reported in a very few cases.

CASE HISTORY

A 31 day female baby born for nonconsanguineous parents was admitted with H/O convulsions (fluttering of eyes and lip smacking movements)-3 episodes 1 day before admission. After admission the child had refractory seizures which was not controlled with Phenobarbitone, phenytoin, levetiracetam and midazolam.

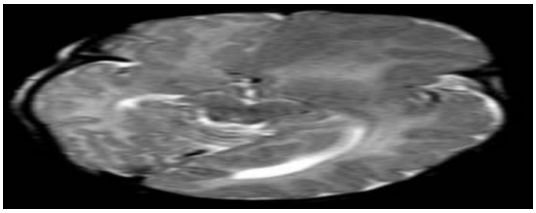


HYPOMELANOTIC LESION CHARACTERISTIC OF HYPOMELANOSIS OF ITO

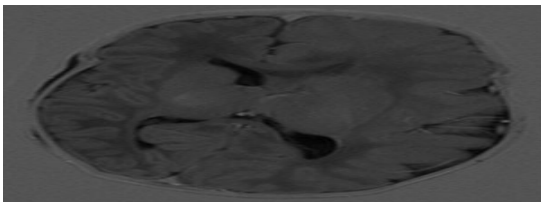
On examination: Head circumference -38.5cm(on 85th percentile of WHO chart), Chest circumference -32.5cm, weight -3.5 kg, length -49cm. *Hypopigmented patch over left temporal region*, AF 2.5*2.5cm,tone in right upper and lower limb and paucity of movements on the same side.

INVESTIGATIONS

Sepsis screen-negative
CSF-normal
Serum electrolytes (Na,K,Ca) -normal
Blood glucose -normal
MRI Brain - Hemimegalencephaly Left cerebral cortex, occipital horn of left lateral ventricle prominent.



HEMIMEGALENCEPHALY OF LEFT CEREBRAL HEMISPHERE



DILATED OCCIPITAL HORN OF LEFT VENTRICLE

DISCUSSION

Hypomelanosis Of Ito is a rare birth defect which presents with characteristic skin lesions. In many cases cause cannot be determined. Usually it is sporadic, in less than 3% family history is positive and appears to be transmitted as autosomal dominant

trait. Autosomal recessive and X linked inheritance has also been reported. The recurrence risk in next offspring depends on the type of inheritance. **Skin lesions**-Hypochromic lesions in distinctive patterns (e.g, whorls, patches, streaks) they often resemble whirled marble. U/L (45%) or B/L, usually show a midline cut off. Patients with hemimegalencephaly have unilateral skin lesions often to contralateral side of brain malformation. In our case lesion is on the ipsilateral side. Skin lesions appear by first year of life in 70% of cases, at birth in 54% of cases.

Central nervous system: Neurological involvement in 76% of cases of patients during first decade of life. Mental retardation and seizures are most common (GTCS 25%,Partial 12% infantile spasms 8% myoclonic seizures 4% out of 50% of patients who presented with seizures). ½ to 2/3 rd have I.Q <70. Somatic hemihypertrophy, macrocephaly or microcephaly may be present. Brain tumors including medulloblastoma and choroid plexus papilloma. Neuropathology-polymicrogyria, disarray of cortical lamination, heterotopic neurons in white matter and giant cells.

HEMIMEGALENCEPHALY-rare congenital disorder of cortical formation with hamartomartous
overgrowth all or a part of cerebral hemisphere.
Incidence-0.1-0.3%of childhood epilepsy.
Clinical features-focal and generalised infantile spasms,developmental delay,hemiparesis and
hemianopia.

EMBRYOLOGY:

The development of cerebral cortex is divided into 3 stages: cell proliferation, cell migration and cortical organization.

Cell proliferation-neuronal and glial precursors are generated, takes place between 2nd and 4th month of gestation. abnormality in this phase leads to too many ,too few or abnormal neurons.

Neuronal migration-from germinal zone to their final destination, close to pial surface in 6 successive waves, takes place during 3rd and 5th month of gestation. Disorders are due to undermigration, overmigration or ectopic migration.

Cortical organization-starts at 22 weeks of gestation till 2nd year of life. Neurons differentiate into several cell types (pyramidal cells, stellate cells) leading to formation of normal cortical cytoarchitectonic pattern. Hemimegalencephaly is a disorder of cell proliferation.

TREATMENT AND PROGNOSIS

Targeted to control seizures. In our case seizures were not controlled with phenobarbitone, phenytoin, levetiracetam and was advised for surgery for which mother refused. Difficult to manage medically. Refractory cases-Hemispherectomy-control in 60% of cases.

FOLLOW UP

The baby continued to have seizures but frequency was reduced with increased doses of antiepileptics.

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