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INFANTILE GM1 GANGLIOSIDOSIS WITH CLASSICAL RADIOLOGICAL FINDINGS- A CASE REPORT

Radiodiagnosis

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ABSTRACT

Infantile GM1 Gangliosidosis is a rare inherited metabolic disorder with an incidence of 1 in 1-2 lakh live births presenting with rapidly progressive neurological and systemic illness associated with skeletal abnormalities, facial dysmorphism , visceral organomegaly, ocular and cutaneous manifestations. We report a genetically proven case of an infant with skeletal, neurological, occular, visceral, cutaneous abnormalities.

KEYWORDS

Infantile GM1 Gangliosidosis, facial dysmorphism.

CASE REPORT:

A 8-month old boy, born via LSCS in view of macrosomia with no significant antenatal history except for polyhydromnios and no perinatal issues presented with global developmental delay, recurrent upper respiratory infections, excessive irritability, abdominal distension and constipation. On examination, the infant was scuporous, extremely irritable and had abdominal distension, round facies with head lag, depressed nasal bridge, downslanting eyes, short broad nose, tented upper lip, Mongolian spots all over body.

Brisk reflexes and augmented startle response to noise were present. No history of seizures, visual and hearing impairment. There was borderline hepatosplenomegaly, bilateral corneal clouding, cherry-red spots. Cardiac evaluation was insignificant.

Radiological findings:

- **D-L Spine**: Dorsolumbar kyphosis, anterior beaking of dorsal and lumbar vertebral bodies with normal bone density.
- **Skull**: J shaped sella with normal skull bones.
- **Hands with distal forearm**: Mild diaphyseal widening of lower ends of radius and ulna.
- **Pelvis:** Flared Iliac bones, shallow Acetabulum, Femoral head was normal.
- **USG Abdomen**: borderline hepatosplenomegaly.
- **MRI Brain**-normal study.

Genetic Analysis:

Homozygous missense variation in GLB1 gene on chromosome 3 was detected.

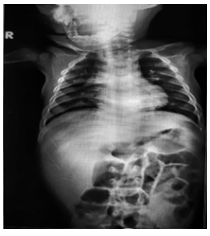

IMAGE -1: AP Radiograph of chest and abdomen showing Hepatomegaly.

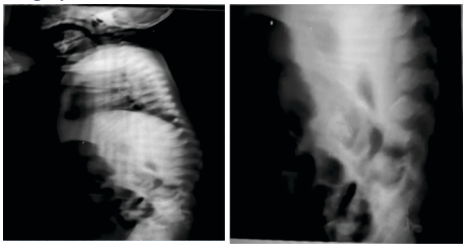

IMAGE-2 & 3: Lateral radiograph of D-L spine and magnified image of Lower lumbar vertebrae showing Anterior spinal beaking.

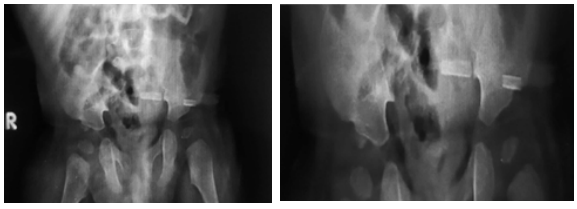

IMAGE-4 & 5: Radiograph of pelvis-AP view & magnified image depicting flared iliac bones, shallow acetabula with normal femoral heads.

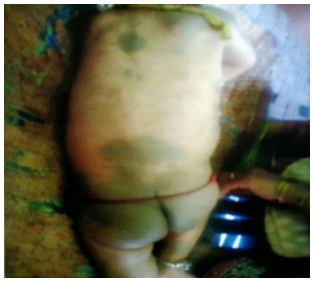

IMAGE-6: Image showing Mongolian spots.

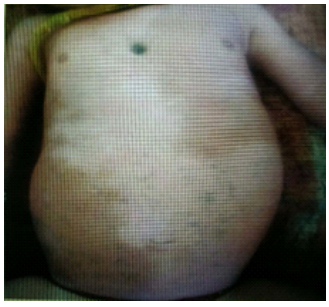

IMAGE-7: Image showing abdominal distension.

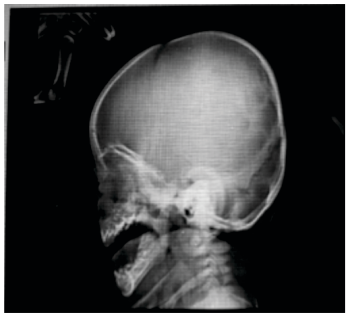

IMAGE-8: Radiograph of skull Lateral view showing J- shaped sella.

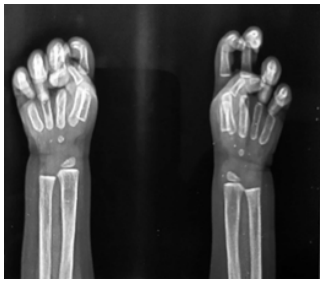

IMAGE-9: AP radiograph of both hands with Distal forearm- mild diaphyseal widening of distal radius and ulna.

DISCUSSION:

GM1 gangliosidosis is a rare autosomal recessive lysosomal storage disorder caused by mutation in GLB1 gene causing a deficiency of betagalactosidase activity and accumulation of gangliosides in various organs [1]. Although it is a rare case, it should be considered as DD for Mucopolysaccharidosis. Three types of GM1 gangliosidosis are reported:

1.**Infantile GM1 gangliosidosis (type 1)**- classic infantile form: is the most common and severe form with affected infants typically appear normal until onset of symptoms but developmental regression can be seen. Signs & symptoms include neuro degeneration, gait abnormalities, seizures, liver and spleen enlargement, coarse facial features, skeletal irregularities, stiffness of joints, muscle weakness, exaggerated startle response to sound. About half of these patients develop cherry red spots in the eye and may become deaf and blind by one year of age[3,4,5]. Rarely patients can have cardiac abnormalities. Neuro imaging findings in patients with Infantile GM1 gangliosidosis have been reported only in few cases.[2] Affected children do not live beyond 2years of age.

2. **Late infantile or juvenile form (type 2)** a less severe form presents between 1-5yrs, with progressive mental and motor retardation, seizures, spastic tetraplegia with cerebellar and extrapyramidal signs. Decerebrate rigidity follows, and death occurs between 3 and 10 years of age, usually precipitated by recurrent bronchopneumonia. Progression of the disease is slower than type 1[6].

3. **Adult or chronic form (type 3)** may have a slow progression with onset between 3 to 30 years of age, presenting with dystonia, dysarthria, ataxia, myoclonus, corneal clouding, gait disorders and extrapyramidal signs. Bony changes are minimal [3,4,5] in this form.

Diagnosis is made by clinical examination, conventional radiography. MRI is required to rule out neurological involvement. However this needs to be confirmed with enzyme analysis of betagalactosidase activity or genetic analysis. Prenatal diagnosis by measurement of enzyme activity in amniotic fluid and cultivated amniotic fluid cells has also been established.

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