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Femoral Hypoplasia Unusual Facies Syndrome: A Rare Case Report

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ABSTRACT

Femoral hypoplasia unusual facies syndrome is a rare form of congenital abnormality characterised by femoral abnormalities and characteristic distinct facial features. Incidence 1 in 25000 births. So far 70 cases have been reported in literature. Association with maternal diabetes has been reported in 35% of cases. Here we report a case of 10 months old male baby second order preterm 32 weeks very low birth weight 1.2 kg EMERGENCY LSCS (Indication - uncontrolled diabetes mellitus with severe oligohydramnios) with left femoral hypoplasia and abnormal craniofacial features.

Keywords: Femoral Hypoplasia, Unusual Facies, Maternal Diabetes Mellitus, Oligohydramnios

CASE HISTORY

A 10 months old male baby of second order birth of non consanguineous parents brought with chief complaints of asymmetry and under development of lower limbs and abnormal facial features.

Baby was preterm 32 weeks delivered by emergency LSCS. Antenatal period mother was a case of un booked pregnancy and did not consume iron and folic acid supplements. Antenatal ultrasound in 32 weeks diagnosed as baby having femoral hypoplasia with severe oligohydramnios. She was admitted in a tertiary care government hospital where she was diagnosed to have uncontrolled diabetes mellitus HBA1C 11 and was started on insulin. Emergency LSCS was done in view of severe oligohydramnios AFI 4 and uncontrolled diabetes mellitus. No history of birth asphyxia or respiratory distress. VLBW-1.2 kg IUGR baby.

Baby was admitted in nicu. Baby had bilateral femoral hypoplasia, sacral agenesis, abnormal facial features. Post natal period was uneventful. Bedside ECHO was normal. Ultrasound abdomen revealed undescended testes with inguinal hernia. Ultrasound cranium Normal. Ophthalmic evaluation was normal. Head circumference was normal 32 cm. Baby was suggested correction of genitourinary abnormalities, orthopedic intervention, and follow up on discharge.

Baby was admitted in paediatric ward at 10 months of age. Baby was evaluated for congenital anomalies. Baby had asymmetrical femoral hypoplasia with left side shorter than right side, bilateral short thighs, sacral agenesis, over riding of toes, Bilateral cryptorchidism, plagiocephaly, low set ears, mongoloid slant, upturned nose, long philtrum (Figure 1-3). Baby had been operated for inguinal hernia at 6 months of age. XRAY B/L hip joint showed bilateral asymmetrical femoral hypoplasia more severe in left side and angulation of femur, poorly formed acetabulum (Figure 4).

MRI BRAIN normal (Figure 5), ECHO was normal.USG abdomen showed undescended testes at level of inguinal ring. Ophthalmic assessment was normal, ENT and Hearing assessment was normal. Child had attained age appropriate milestones as per corrected gestational age.

Baby was advised orchidopexy. Orthopedic opinion obtained advised follow up and orthopedic aid as child grows. Baby was suggested early stimulation and regular follow up

Figure 1

Figure 2

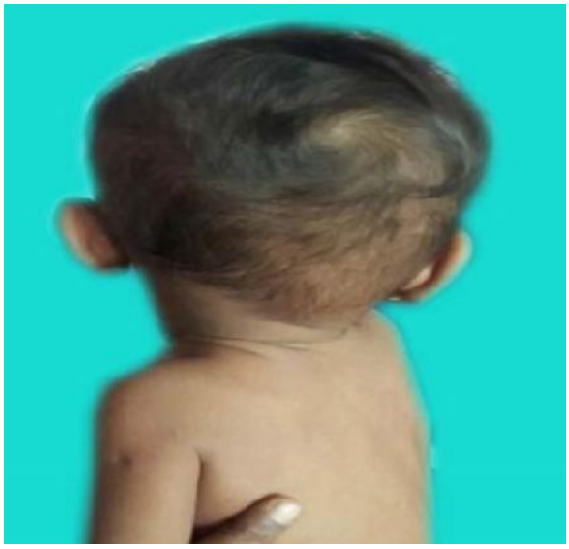


Figure 3

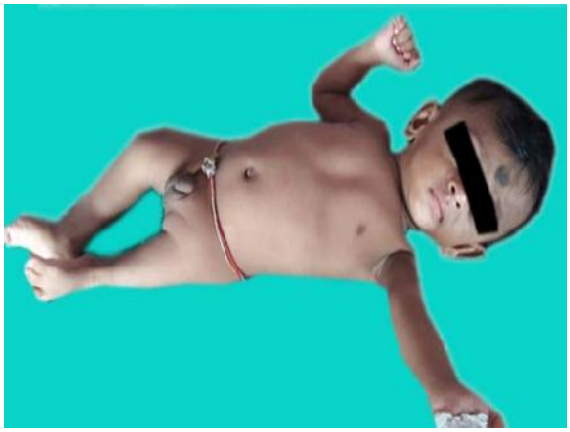


Figure 4: Xray hip-bilateral asymmetrical femoral hypoplasia left side more severe than the right side, Lateral bowing of femur in right side

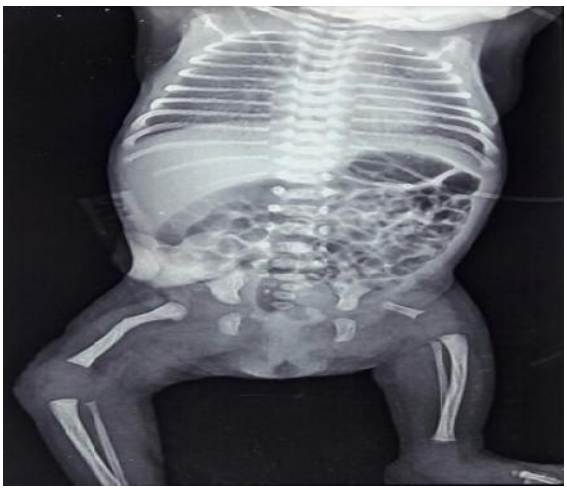
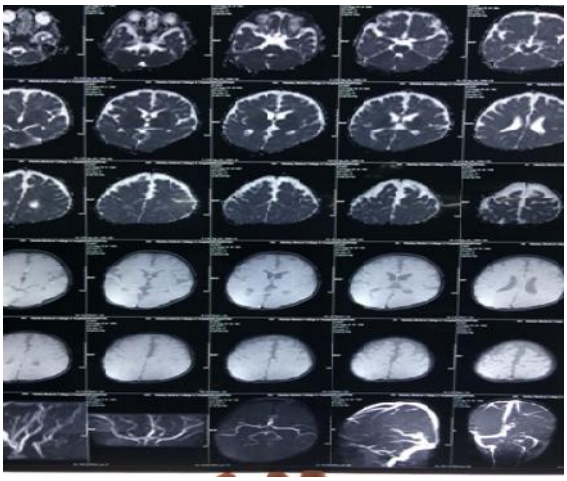


Figure 5 : MRI brain of this child-normal



DISCUSSION

Femoral facial syndrome (FFS) or Femoral hypoplasia unusual facies syndrome (FHUFS) is a rare syndrome first reported in 1961 by Franz Ch and O''Rahilly R consisting of asymmetrical femoral hypoplasia and abnormal facial features. Further case reports were submitted by Jones KL et al and Daentl et al. FHUFS syndrome closely resembles caudal dysplasia syndrome or sirinomyelia which occurs due to insufficient mesoderm in the caudal part of embryo causing lumbosacral defects, lower limb dysplasia, renal agenesis but facial abnormalities are absent. Most cases are sporadic or multifactorial in etiology. one case of autosomal dominant inheritance has been reported. Association with maternal diabetes reported in 35% of cases. It is 200-300 times more common in IDM mothers. Disruption of normal carbohydrate homeostasis at a critical point of embryogenesis is responsible for FHUFS. Maternal diabetes is a teratogenic factor.

ABNORMALITIES

Growth: short stature due to short lower limbs

Craniofacial abnormalities: plagiocephaly, low set ears (30 -80%), upturned nose, long philtrum, thin upper lip, cleft lip cleft palate, micrognathia, mongoloid slant

Limbs: bilateral usually asymmetrical hypoplastic femur (80-99%), sacral agenesis (30-80%), flat buttocks, Dysplastic hips (30-80%), flexion and adduction deformity of hips, overriding of toes, talipusequinovarus.
Pelvis:hypoplasticacetabulae

Spine: Dysplastic sacrum,scoliosis, hemivertebrae

Genitourinary: cryptorchidism, inguinal hernia, micropenis, polycystic or dysplastic kidneys, hypoplastic labia.

CNS: Corpus colossal agenesis

Occasional Abnormalities: septal defects like VSD, pulmonary stenosis, truncusarteriosus, craniosynostosis, preaxialpolydactyly of feet, astigmatism, fused or missing ribs

Though there may be problems in speech development, the patients are of normal intelligence. most of them are ambulatory.

CONCLUSION

Femoral hypoplasia unusalfacies syndrome is a rare syndrome which may be associated with maternal diabetes or oligohydramnios. In our case it is a form of diabetic embryopathy with oligohydramnios.pre pregnancy control of diabetes and control of gestational diabetes becomes very important to avoid such birth anomalies. Antenatal ultrasound is very useful to detect femoral hypoplasia and micrognathia. Before foetal viability the option of termination of pregnancy can be discussed with parents. Post natal management is directed towards correction of genitourinary, cardiac, facial abnormalities. Orthopedic aids should be provided to such children. Regular follow up, immunization and growth monitoring is necessary. Family counseling and planning of subsequent pregnancies should be given to parents.

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