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GREENBERG'S DYSPLASIA- A RARE GENETIC CONDITION

Pediatrics

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KEYWORDS

INTRODUCTION:

Greenberg's Dysplasia, also known as Hydrops-Ectopic calcification-Moth-Eaten (HEM) Skeletal Dysplasia, is a rare autosomal recessive osteochondrodysplasia, caused by mutation in the Lamin B Receptor (LBR) Gene, on chromosome 1q42.

Case History:

23 year old primigravida, with h/o consanguineous marriage, came for routine antenatal check up at 17 weeks of gestation. USG was done which demonstrared left foot- multiple cystic hygroma, short limbs depicting short humerus and one forearm bone. Irregular hypo- and hyperechogenic foci were observed in the bones. Pericardial effusion was present. These features were suggestive of Greenbergs's Dysplasia.

Management:

Because of the severity of the lethal condition, the parents were counselled and MTP was performed at 20 weeks of gestation, after obtaining consent.

- The weight of the fetus was 447 grams.
- Extensive Swelling of the Body (Hydrops Fetalis).
- The fetus had a large head with depressed nasal bridge and short Barrel shaped trunk.
- Short limb Dwarfism (Micromelia) and Polydactyly.

Radiological Abnormalities included ectopic calcification of long bones, ribs, pelvis and disorganisation of chondroosseous calcification along with Platyspondyly.

Histopathological Changes include chondrocytes with inclusion bodies with homogeneous material, along with disorganization of cartilaginous architecture.

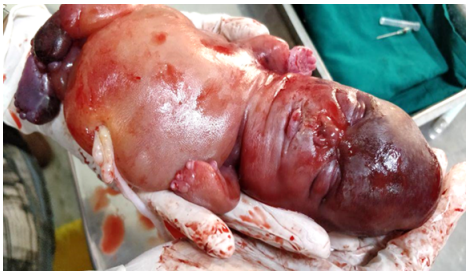
DISCUSSION:

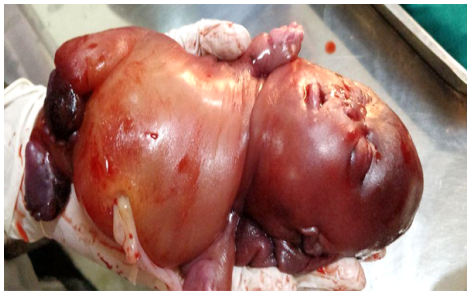
Greenberg dysplasia is a severe condition characterized by specific bone abnormalities in the developing fetus. In the year 1988, Greenberg et al. described 2 siblings, the offspring of consanguineous parents, who presented with an apparently 'new' severe form of short-limb dwarfism. The chondroosseus radiologic and histologic features were distinctive. The first sib presented at 30 weeks of gestation with severe hydrops following fetal death; the second was detected by ultrasonography at 20 weeks. Radiologic abnormalities included an unusual 'moth-eaten' appearance of the markedly short long bones, bizarre ectopic ossification centers, and marked platyspondyly with unusual ossification centers. Extensive extramedullary erythropoiesis was found in both fetusesThe bones of affected individuals do not develop properly, causing a distinctive spotted appearance called moth-eaten bone, which is visible on x-ray images. This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. Mutations in the LBR gene cause Greenberg dysplasia. This gene provides instructions for making a protein called the lamin B receptor. One region of this protein, called the sterol reductase domain, plays an important role in the production (synthesis) of cholesterol. *LBR* gene mutations involved in Greenberg dysplasia lead to loss of the sterol reductase function of the lamin B receptor, and research suggests that this loss causes the condition. Absence of the sterol reductase function disrupts the normal synthesis of cholesterol within cells. This absence may also allow potentially toxic byproducts of cholesterol synthesis to build up in the body's tissues. It is not known, however, how a disturbance of cholesterol synthesis leads to the specific features of Greenberg dysplasia. Greenberg dysplasia is a very severe condition that causes a fetus to not survive to birth.

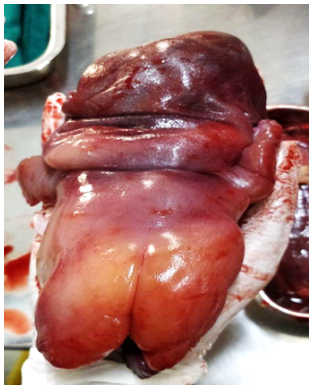
CONCLUSION:

Greenberg's dysplasia is a very lethal condition, that caused the fetus to not survive until birth. There are currently less than ten published cases in literature. Genetic counselling plays an importance role. There is evidence in literature stating that this condition can affect recurrent pregnancies too and unfortunately no means of preconceptional diagnosis is available.

Image Gallery:







The above images depict the fetus demonstrating hydrops fetalis.

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