Case report
Inborn error of metabolism
Smith-lemli opitz syndrome

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1day-Old Child First in Birth Order born of 1st degree consanguineous marriage presented with Respiratory distress syndrome

He was born PT 30weeks, breech delivered with birth weight of 1.8kg appropriate for gestational age. Cried after 5 minute of birth.
Antenatal history: mother had primary infertility of 8years.

On examination, weight was 1.8kg, length 40cm, and HC was 32cm.He had narrow frontal area, low set ear, inner epicantal fold, broad nasal tip with ante verted nostril, micrognathia, simian crease, and ambiguous genitalia : micropenis, hypospadiasis, cryptorchidism, and rudimentary vagina, and severe respiratory distress baby expired within 1 hour due to acute respiratory failure. Diagnosis was made on the clinical background

Smith-Lemli-Opitz syndrome (SLOS) is a multiple congenital anomalies/mental retardation (MCA/MR) syndrome is rare autosomal
A recessive disorder of cholesterol metabolism where in the conversion of 7-dehydro-cholesterol into cholesterol is disrupted leading to excessive accumulations of 7-DHC, 8-DHC and cholesterol deficiency. Mutations in the DHCR7 gene are responsible for SLOS. Spontaneous abortion, congenital heart diseases, death due to multiorgan failure in 1st week, failure to thrive, vomiting, constipation, hearing and visual loss may be present.

Diagnosis was made based on clinical and biochemical profile like low cholesterol and absent LDL cholesterol by gas-liquid chromatography or gas chromatography or by tandem mass spectrometry, karyotype, and ultrasound abdomen. Hence, cholesterol replacement forms the cornerstone in medical management and coexisting surgical condition may need appropriate evaluation and intervention.

Prenatal diagnosis by foetal ultrasound may reveal multiple congenital anomalies, increased 7-DHC and mutation analysis in amniotic fluid, or chorionic venous sampling, decrease maternal serum unconjugated estriol or other marker for triple test with normal karyotype suggestive of SLOS. The unique presence of equine estriols in the maternal urine during pregnancy of a fetus affected by SLOS, potentially allowing noninvasive prenatal screening for SLOS.

Photograph attached with this copy

Reference:


