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Case Report Open Access

A Case with Congenital Cataract, Ascitis, Ambiguous Genitilia with Hydronephrosis with Micrognathia

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Abstract

A case with multiple congenital anomalies is very challenging to diagnose. We always has to keep some differential diagnosis. Here we are reporting a case with multiple congenital anomalies (congenital cataract, ascitis, micrognathia, ambiguous genitilia). It can be Smith-Lemli-Optiz Syndrome (SOLS) or syndrome or Gardner-Silengo-Wachtel syndrome. Aim of presentation is awareness of these syndrome. Some syndromes like Smith-lemli-Optiz syndrome it can be detected in pre-natal period and can be treated with cholesterol supplementation.

Keywords: Cataract; Ambiguous genitilia; Syndrome; Congenital

Case Report

We are describing a case of 19 Year unbooked female primigravida at 33+6 weeks of gestation reported to labour room with pain abdomen and leaking per vagina. On examination her pulse was 86 per minute. Blood pressure was 110/70. On abdominal examination single live fetus, term size and fetal heart sound 140/minute. Uterine contractions present. On per vagnal examination cervix was 5 cm dialated and fully effaced. During antenatal period she received two doses of tetanus toxoid. She has an USG which was showing bilateral hydroureternephrosis, dialated bowel loops. She delivered vaginally a single live baby with congenital cataract, ambiguous genitilia and ascitis. Baby was having bilateral hydro-utretero-nephrosis, micrognathia. Size of left eye is less than right. Birth-weight-weight-2400 gm. Baby was having respiratory distress and admitted in NICU and intubated and kept on ventilator. But the condition of baby deteriorated and expired after 24 hrs. The blood of baby was sent for basic investigations (hemoglobin, TLC, DLC, Sugar, Blood Grouping & Karyotpe). Karyotpe shows 46XY. We could not diagnose this case. We search the literature and made the following differential diagnosis (Figures 1-3).

Differential Diagnosis

1. Smith-Lemli-Optiz Syndrome (Microcephaly, mental retardation, brain anomalies, cataract, micro-opthalmia, ptosis, cleft lip/palate, pyloric stenosis, Hirschsprng disease, Congenital heart defect, renal anomalies (hypoplasia, hydronephrosis), Genital anomalies, Polydactyly) [1,2].

2. Gardner-Silengo-Wachtel syndrome (Micrognathia, low set ears, double outlet right ventricle with VSD and 46 XY Gonadal dysgenesis). No polydactyly (Figure 4).

3. Ullrich-Feichtiger syndrome

4. Meckel –Gruber syndrome

5. Noonan syndrome (autosomal dominant)

6. Pseudotrisomy 13 syndrome (holoprosencephaly-polydactyly)

In this case we have not sent 7-DHC level.

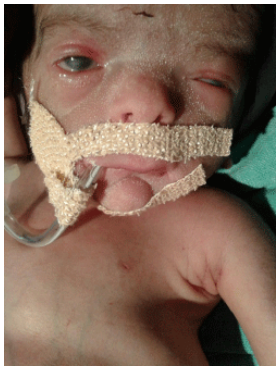

Figure 1: Congenital cataract

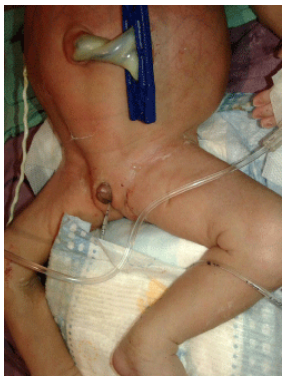

Figure 2: Ambiguous genitilia

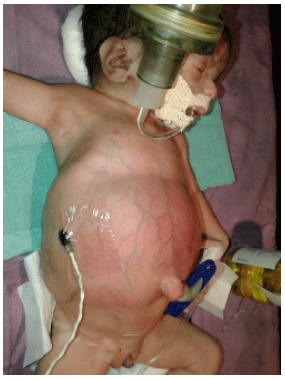

Figure 3: Ascitis

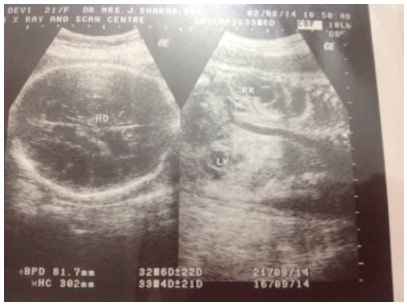

Figure 4: Hydronephrosis

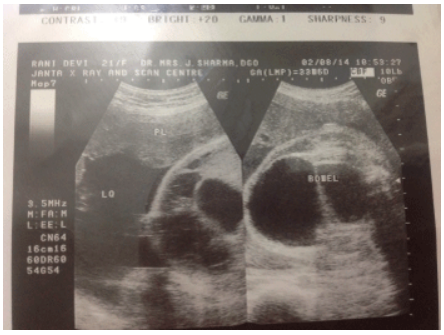

Figure 5: Dialated bowel loop

Discussion

In this case of multiple congenital anmalies clinically we cannot come to a diagnosis. So we have keep various differential diagnosis in mind. Clinical features of this case resembles either with Smith-lemli-Opti syndrome or Gardner-Silengo-Wachtel syndrome [3-5].

Smith-Lemli-Optiz syndrome (OMIM-270400) is an autosomal recessive disorder caused by mutation in gene encoding 7DHCR which is located on chromosome 11q13. Prevalence is 1 in 20000 to 1 in 60000 More in Caucasians. SLOS is characterized by faulty cholesterol synthesis which will cause mental retardation, growth restriction and various congenital anomalies of genito-urinary and gastro-intestinal tract (Figure 5).

Clinical Features

Microcephaly, mental retardation, brain anomalies, cataract, micro-opthalmia, ptosis, cleft lip/palate, pyloric stenosis, Hirschsprng disease, Congenital heart defect, renal anomalies (hypoplasia, hydronephrosis), Genital anomalies, Polydactyly.

Diagnosis

1. Biochemical- Elevated 7 DHC levels which are measured in plasma or tissues.

2. DHRC7 mutation analysis can be done.

3. Prenatal Diagnosis

- Low maternal serum levels of unconjugated oestriol

- Measuring 7DHC levels in amniotic fluid or CVS

- Direct mutation analysis of DNA isolated from amniocytes or CVS

- Treatment is cholesterol supplementation.

4. In Gardner-Silengo-Wachtel syndrome (Micrognathia, low set ears, double outlet right ventricle with VSD and 46 XY Gonadal dysgenesis). No polydactyly.

Conclusion

Whenever a baby with multiple congenital anomalies with ambiguous genitila presents we should keep various syndromes like SLOS Syndrome in mind. It can be detected in prenatal period. It can be treated with cholesterol supplementation. If we had detected prenatally this baby could have been saved.

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