



A Rare Case: Pregnancy with Robert's Syndrome Foetus

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Introduction

Robert's syndrome is a rare genetic disorder characterized by prenatal and postnatal growth retardation, limbs, and craniofacial defects. The limb defects are similar to those seen in Thalidomide embryopathy, hence, the disorder is also known as pseudothalidomide syndrome.

Robert's syndrome occurs equally in both male and female baby and is common among closely related parents (parental consanguinity). Only about 150 cases have been described in the literature in the world over.

Aim

To present a rare case of pregnancy with Robert's Syndrome baby with polyhydramnios who underwent normal vaginal delivery.

Place of study

Department of Obstetrics and Gynecology, Katihar Medical College and Hospital, Katihar

Case Study

A 26 year primigravida with 7 and half months' amenorrhea presented in obstetrics casualty with chief complains of pain abdomen since 1 day.

Pain was sharp aching, radiating to back and not relieved by taking medication. Mild shortness of breath was present due to polyhydramnios.

On Examination

BP= 110/70 mm of hg

P/R= 92 bpm

Temp= 98 F

Pallor= +

Per Abdomen- uterus unduly enlarged, head-engaged, FHS not localized by stethoscope or CTG

Per eye- No active leaking or bleeding pv

Per Vaginal – cervical os 3- 4cm dilated, membrane present

Investigation

Hb- 9.8

Platelets- 91000

ABoRh- A positive

TLC- 5600

RBS- 110

S. Urea- 28

S. Creatinine- 0.84

SGOT- 30

SGPT- 42

S. ALP- 94

S. Bilirubin Total- 0.80

Direct- 0.40

Indirect- 0.40

HIV- NR

HBsAg- NR

Anti HCV- NR

USG (lower abdomen)- Single non-viable foetus of 29 weeks 4 days, with all limbs severely small (S/O severe macromelia), massive polyhydramnios, minimal hydrocephalus, cleft lip.

Operative Procedure

After all preliminary investigations, induction of labour was done. Under Aseptic and antiseptic precaution, patient laid in lithotomy position, perineal parts painted and draped. Artificial rupture of membrane was done. Gush of massive amount about 2litres litres of amniotic fluid came out. Patient asked to bear down with each uterine contraction. IUD male foetus with weight 450 grams, was delivered on 17/06/21. Cord clamped and cut. Placenta and membrane taken out completely. Uterine cavity explored. Hemostasis achieved. No tear or PPH occurred. Betadine vaginal toileting done.

Post Delivery

She was given injectable antibiotics and iron and calcium supplements. The postpartum period of the mother was uneventful. She was discharged from the hospital after 24 hours. She was advised for pre conceptional counselling to be done in next pregnancy and take proper ANC visits in next pregnancy.



Discussion

Robertson's syndrome is a rare autosomal recessive condition. An important aspect to consider is the variable expression of the syndrome. This variability affects both the phenotype and the genotype. Robertson's syndrome can, infact be diagnosed on cytogenic grounds in the absence of major congenital anomalies. The presence of mid-facial clefts (lip and palate) nose and ear abnormalities, facial hemangiomas, hypertelorism with prominent eyes and corneal clouding, microcephaly, symmetric limb abnormality, severe growth and mental retardation are very suggestive of Robert's syndrome. Less common findings are oligo dactyly, micrognathia, cryptorchidism, oligohydramnios, renal anomalies (polycystic or dysplastic kidney) and heart defects (in particular atrial septal defect and patent ductus arteriosus). In our case study, Robert's syndrome was suspected only on the basis of the ultrasonographic detection of the complex of

multiple malformation, since the couple had a negative family history. There is 25% recurrence risk due to autosomal recessive transmission, thus the perinatal diagnosis of this syndrome is important for couple with a positive family history.

In these cases, a chorion biopsy at 8-10 weeks followed by a transvaginal scan at 12-14 weeks might represent the procedure of choice. When detected before viability, termination of pregnancy can be offered. After viability, standard obstetrical management is not altered.

Because of the signs of the disorder are similar to those caused by the ingestion of Thalidomide by pregnant women, the term Pseudo-thalidomide, is frequently used.

Finally, prenatal diagnosis for at risk pregnancies is recommended and requires either prior identification of the disease causing mutation in the family or ultrasound examination combined with cytogenetic testing.

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