

DECOMPRESSION CRANIOTOMY FOR SHORT RIB POLYDACTYLY SYNDROME: A CASE REPORT

Presented by:

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Objective



The World Health Organization (WHO) recommends one obstetric ultrasound (US) before 24 weeks gestation to confirm gestation and rule out anomalies. In our setup, we encounter term parturients with no previous US. Despite the low incidence of congenital anomalies, missed diagnoses may increase peripartum morbidity and mortality. We report the case of short rib polydactyly syndrome (SRPS) in late second stage with an entrapped after-coming hydrocephalic head.

Case Report



A 25-year-old gravida 4, para 2+1 presented with a dead fetus dangling from the introitus with an entrapped head. Maternal exhaustion was noted.

Mauriceau-Smellie-Veit (MSV) maneuver was unsuccessful. A bedside US revealed a massive hydrocephalus and a fundal placenta. Informed consent for decompression craniotomy was obtained from the mother.

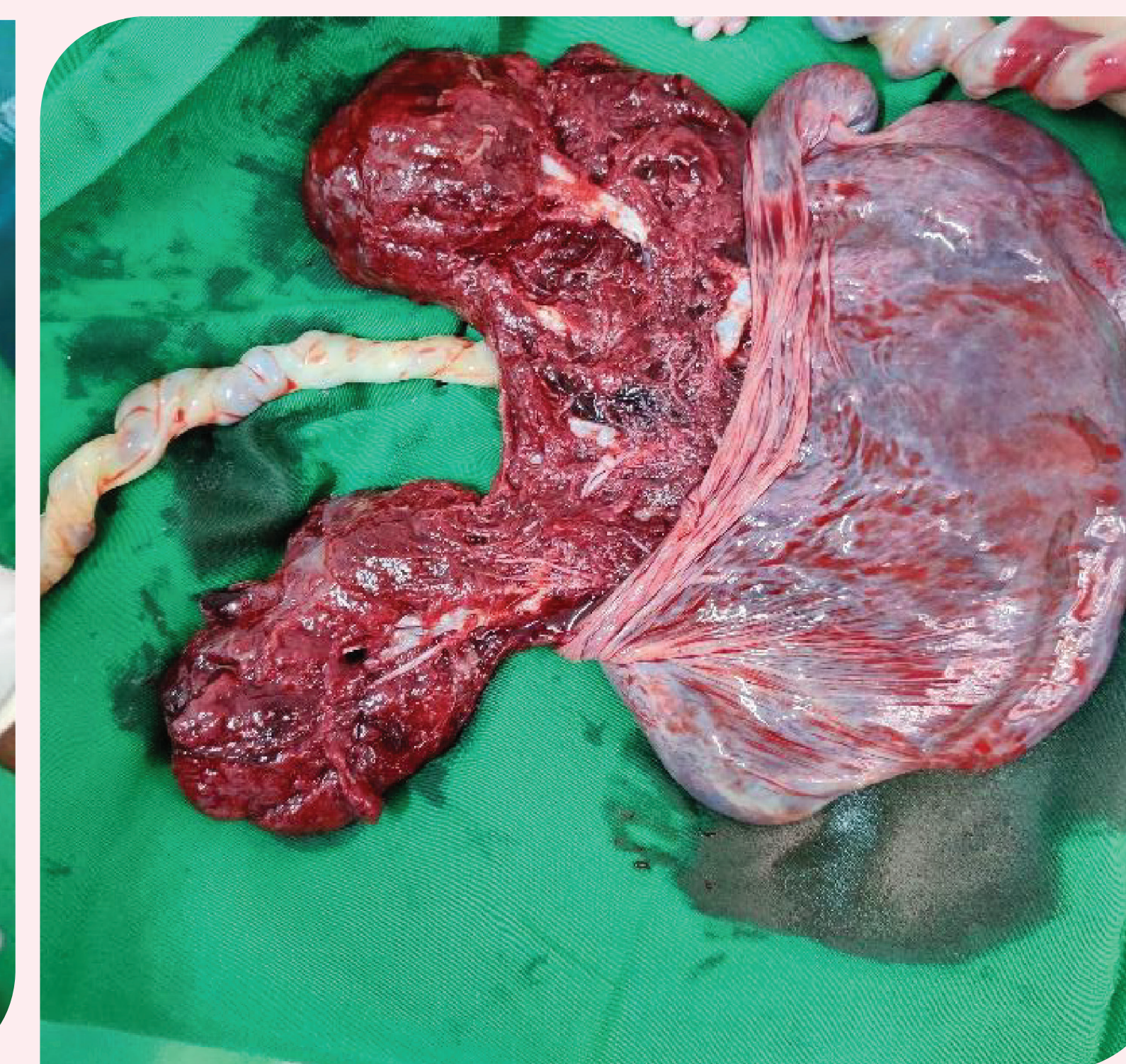
Spinal anesthesia was administered and bladder emptied aseptically.



The posterior fontanelle was not palpable. A 5mm bariatric-use laparoscopic trocar was introduced via the foramen magnum of the fetus under US guidance. Clear cerebrospinal fluid (CSF) was drained. The decompressed head was then delivered using MSV maneuver. Active management of the third stage of labor was administered.

No major perineal injuries were noted. Blood loss was minimal. The patient recovered and was discharged home after 24 hours.

Postmortem revealed a 2295-gram baby with an enlarged head (circumference 38 centimeters) punctured posteriorly. Chest circumference was 28cm. The abdominal girth was 31cm. Gross external anomalies included a cleft lip-palate, underdeveloped eyes, low set underdeveloped ears, micromelia, polydactyly of upper and lower limbs, and ambiguous genitalia. The anus was patent. The parents declined the dissection of the baby. A diagnosis of SRPS was made.



Discussion

SRPS is an autosomal recessive disorder with an incidence of 1/500,000 births and 25% recurrence rate. There are 4 types, and types I and IV spectrums were present in this baby. Elective termination of pregnancy is recommended when diagnosis is made antenatally. Our patient's first US was done on the day preterm labor begun, revealing a single live intra-uterine pregnancy at 36-weeks gestation with holoprosencephaly and multiple malformations. The late diagnosis made management difficult.

Genetic testing is recommended but patients from low-and-middle-income countries rarely afford these tests.

Conclusion

The need for early anomaly US in pregnancy should be overemphasized. This allows for apt prenatal diagnosis and management, preventing unnecessary physical and emotional trauma to mothers and caregivers during pregnancy and childbirth.