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A case of Fraser syndrome

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Objective

To describe a case of Fraser syndrome differentiated from Manitoba oculotrichonal (MOTA) syndrome in the postnatal period according to the morphological features.

Methods

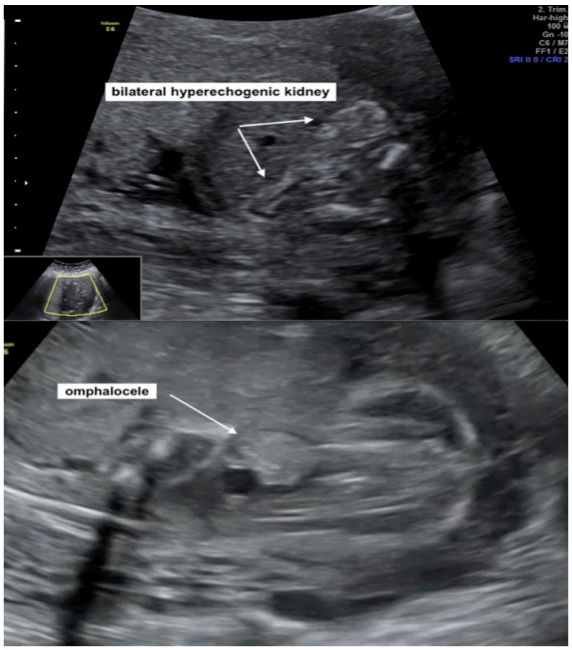
The antenatal record of a case of postnatally diagnosed Fraser syndrome was reviewed retrospectively.

Results

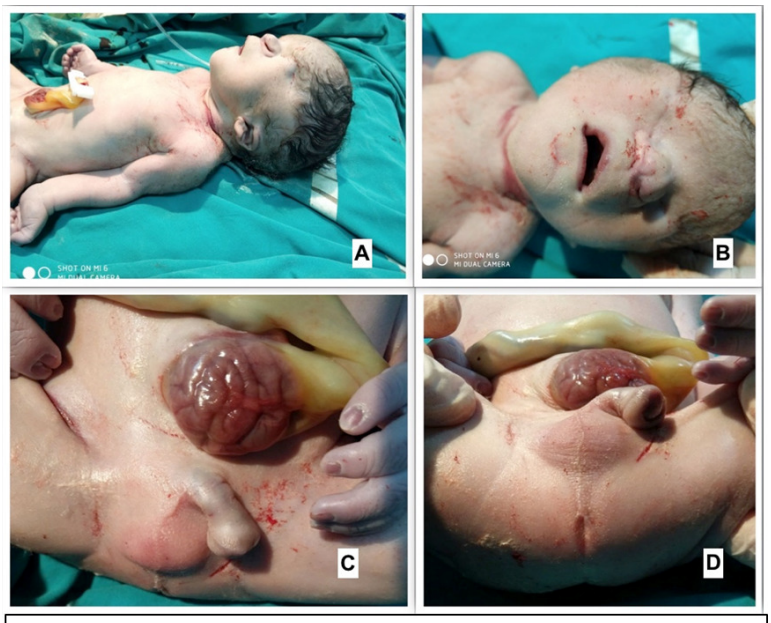
A 27-year-old pregnant woman with a history of one first-trimester miscarriage was referred to our Prenatal Diagnosis and Treatment Unit for second-trimester detailed ultrasound examination due to oligohydramnios at 22 weeks gestation. Her medical history was unremarkable. Physical examination and biochemistry were normal. Obstetric ultrasound revealed that the fetal biometry was compatible with gestational age and the placenta was located on the anterior wall of the uterus with a normal appearance. The amniotic fluid index was 1. 4 cm suggesting severe oligohydramnios. Bilateral hyperechogenic kidneys and an abdominal wall defect (omphalocele) were detected in the fetal anatomical assessment. (Picture-1). The option of invasive prenatal diagnosis was offered, which the parents refused, and subsequent follow-up scans were performed at two weekly intervals. Elective cesarean section was planned at 34 weeks of gestation due to anhydramnios and intrauterine growth retardation after one course of betamethasone administration. A male infant was delivered, Apgar scores were 3 at one minute and 4 at five minutes, birthweight was 2100 g, length was 47 cm. On physical examination, he had a bilateral wedge-shaped anterior hairline from the temporal region to the ipsilateral eye, anophthalmia, omphalocele and anal atresia that suggested MOTA syndrome (Picture-2). Neonatal intubation could not be performed in the neonatal resuscitation effort which was compatible with laryngeal/tracheal atresia. There was also syndactyly in both hands and feet (Picture-3). Fraser syndrome was considered in the differential diagnosis. The baby did not respond to neonatal resuscitation, and died in the first hour of life. Postmortem autopsy and genetic testing was offered to the parents, but unfortunately, the parents also refused these.

Conclusion

Phenotypic similarity between Fraser syndrome and MOTA syndrome includes cryptophthalmos, anophthalmia/microphthalmia, a wedge-shaped temporal anterior hairline, hypertelorism, a bifid nasal tip or notched nares and imperforate anus. Both syndromes have an autosomal recessive inheritance and are more seen in consanguineous parents. However, babies with MOTA syndrome have not had syndactyly, ambiguous genitalia, cognitive disease, ear or limb anomalies. MOTA syndrome is compatible with life, and cognitive development is usually normal. Besides, early mortality is frequently observed in individuals with Fraser syndrome. The case is presented because of its rare occurrence and postnatal detection on the basis of clinical findings.



Picture 1. Ultrasonographic image of bilateral hyperechogenic kidney and abdominal wall defect (omphalocele).



Picture 2. The demonstration of bilateral wedge-shaped anterior hairline from the temporal region to the ipsilateral eye (A), anophthalmia (B), omphalocele (C) and anal atresia (D) in the neonate.