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Meckel-Gruber Syndrome in One of Nonidentical Twins: Short Case Report

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A male twin pair, the result of 34 weeks gestation by dates, 38 weeks by estimation, was delivered at our hospital by normal vaginal delivery on 02.01.87.

The mother is a 27-year old, A,Rh+ healthy school teacher. She was married to her 33-year old, first-degree cousin in 1981. They have no family history of malformed newborns or other inherited diseases.

The first pregnancy of this gravida-four mother was in 1982 and resulted in a full-term, healthy female weighing 3.5 kg. The second pregnancy was in 1984 and resulted in a 36-weeks, alive female with occipital encephalocele, who died 12 hr after birth. It is not known if this newborn had other accompanying congenital anomalies. The third pregnancy was in 1985, and the fetus was aborted at 12 weeks gestation because of anencephaly, which was diagnosed by ultrasound.

The last pregnancy was followed at a private clinic without doing ultrasound. It was not complicated by any illness, drug intake, or X-ray exposure.

After 34 weeks of gestation, male twins were delivered by spontaneous vaginal delivery. Each one had his own placenta and umbilical cord. The first twin was a healthy male weighing 2.4 kg, with Apgar score 7/8, who was discharged home in a good general condition, and was doing well when he was seen at follow up one month later.

The second twin was a male weighing 2.15 kg, and having a length of 48 cm. We was microcephalic with a head circumference of 28 cm.

Examination of the head showed an occipital ruptured encephalocele measuring 5 cm in diameter, low-set ears, and a sloping forehead. His anterior fontanel was wide (6 x 4 cm) with separation of suture lines.

Brain ultrasound showed dilated lateral ventricles with intracerebral hematoma. He had a short neck and a narrow bell-shaped thoracic cage. The umbilical cord showed two arteries and one vein with no omphalocele. Abdominal examination revealed bilateral

flank masses measuring 7 x 4 x 3 cm each, which were proved to be bilateral polycystic kidneys by abdominal ultrasound. There was also a right reducible inguinal hernia. He had normal male genitalia with bilateral descended testes.

Extremities showed arthrogryposis with bilateral talipes equinovarus. No polydactyly, syndactyly or simian creases were seen.

He was diagnosed as a case of Meckel-Gruber syndrome, which is inherited as autosomal recessive. Detailed blood grouping was done for both twins and proved that they were nonidentical twins. The affected twin died two days after birth.

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