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MECKEL SYNDROME IN TWINS*

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Meckel syndrome was first described in two siblings by Meckel in 1822[1]. In 1934, Gruber coined the term "dysencephalia splanchnocystica" to describe the malformations associated with this syndrome[1]. In 1969, Opitz and Howe reported the first case in the English literature and suggested the term Meckel syndrome[2]. In 1980, Tunçbilek et al[3] reported five cases of Meckel syndrome in Turkey.

The main defect in this syndrome, which is inherited as an autosomal recessive trait, is not known. The basic characteristics of this syndrome are encephalocele, polycystic kidneys and polydactyly, and in order for a diagnosis of Meckel syndrome to be made, two of the afore-mentioned features are required.

In this paper, we present Meckel syndrome in twins in order to emphasize the importance of genetic counselling, since prenatal diagnosis is possible.

Case Report

A 33-day-old male infant, who was operated on for encephalocele and had hydrocephalus, was transferred to our clinic. He and his twin sister were born with encephaloceles and were operated on when they were three-days-old. The materials which were extirpated were examined in the Pathology Department of our University Hospital. The twin sister died postoperatively.

The mother, aged 28 years, and father aged 40 years, were second degree relatives. There was also an 11-year-old sister and a nine-year-old brother, who were healthy.

Physical examination revealed a hypoactive, hypothermic infant. Sucking was absent. His weight was 3150 g, length 52.5 cm, head circumference 39.5 cm,

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pulse 96 per min and body temperature 35.6°C. All the cranial sutures were separated and the anterior fontanelle was 4 × 6 cm. The eyes appeared to be deviated downward (setting-sun sign), leukoma was present in the left eye and micrognathia was evident (Fig. 1). The liver was enlarged two cm. Abdominal masses, possibly of the kidney, were palpated. One was 7 × 7 cm and localized in the right lower quadrant, and the other 7 × 5 cm and localized in the left lower quadrant.

Laboratory studies revealed that the hemoglobin level was 9.1 g/dl, white blood cell count 9,100/mm^2, with a distribution of 70% neutrophils and 30% lymphocytes. The morphology of the RBCs were normal and platelets were adequate. Urinalysis yielded an acid reaction, density 1004, protein and glucose were not present. The blood chemistry showed blood glucose 73 mg/dl, blood urea nitrogen 19 mg/dl, Na^+137 mEq/lt, K^+3.8 mEq/lt, and serum calcium 11 mg/dl. Serological tests for cytomegalovirus, toxoplasmosis and rubella were all negative. Amino acids in the blood and urine were normal. Chromosome analysis was also normal.

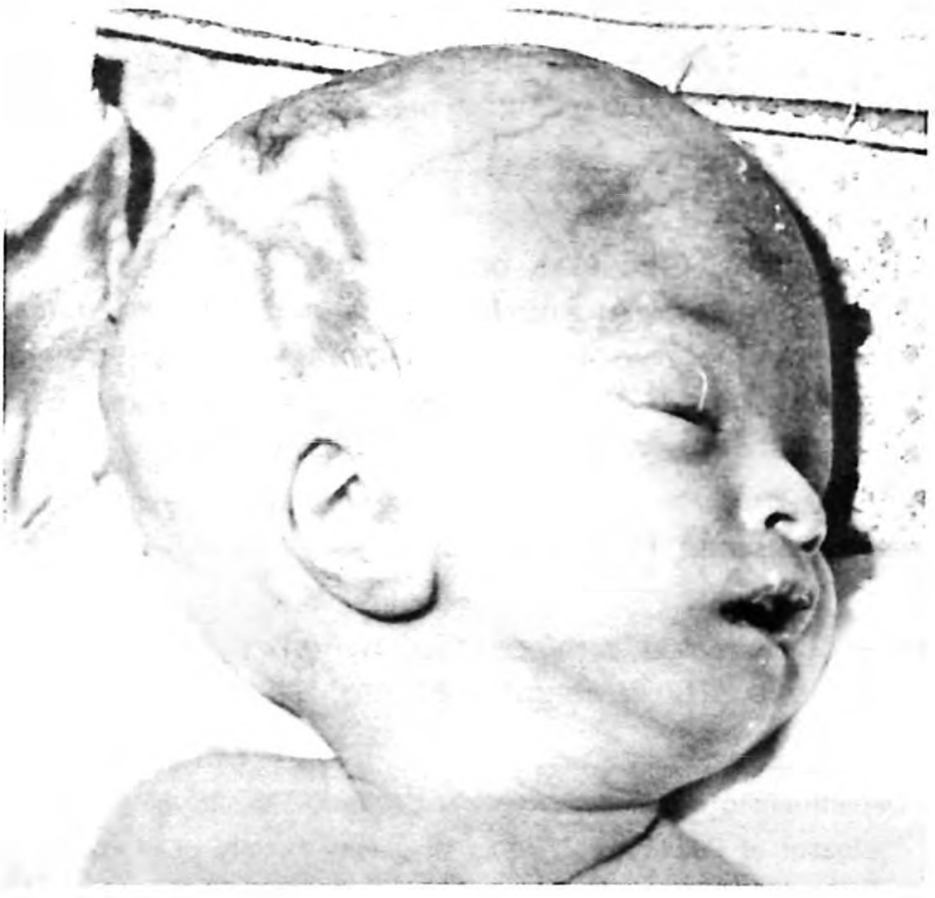

Fig. 1: The appearance of the face, hydrocephalus and micrognathia.

Computerized tomography of the brain revealed thin cortical tissue and abnormal enlargement of the ventricular structures while computerized tomography of the abdomen showed polycystic kidneys (Fig. 2). The encephalocele sac was covered with hairy skin. Microscopically meningeal and brain tissues were reported among the epithelial structures. Postmortem examination revealed polycystic kidneys.

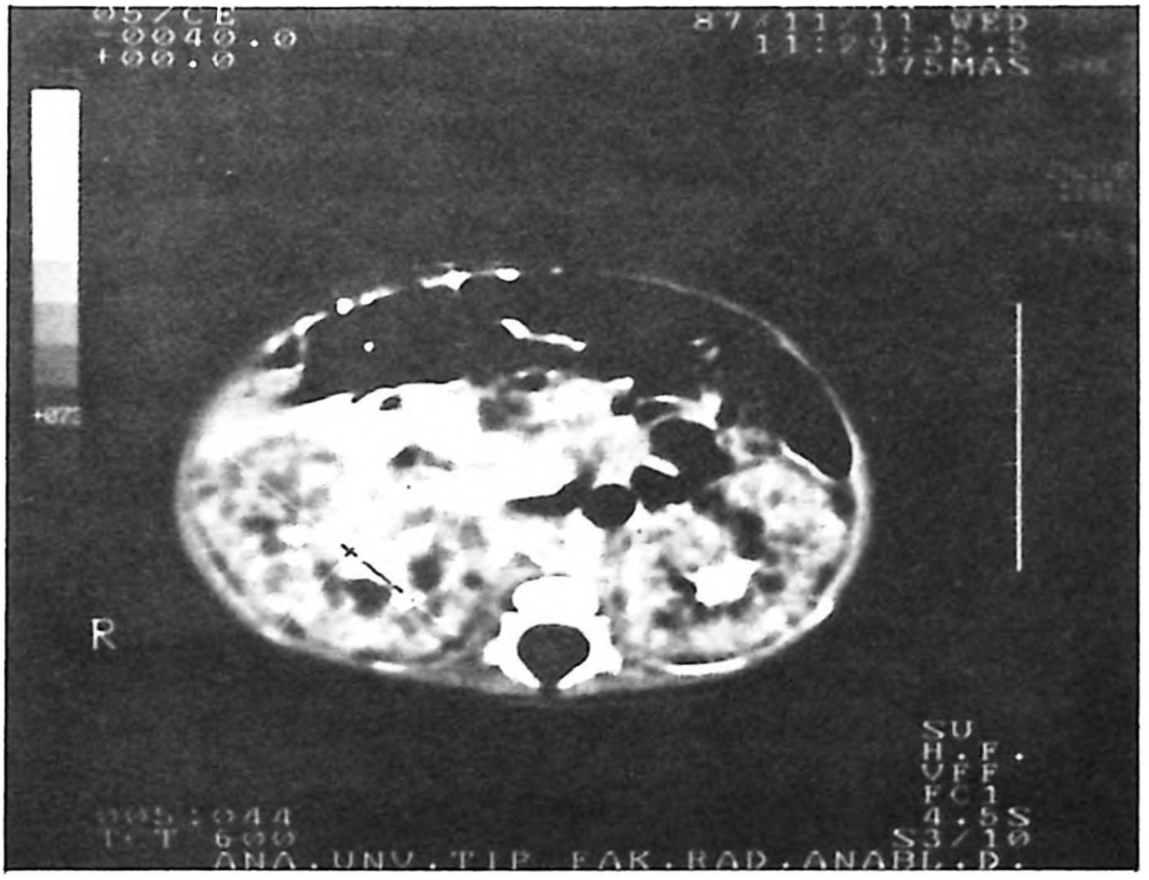

Fig. 2: Abdominal computerized tomogram; polycystic kidneys.

Discussion

The most striking findings in our case were encephalocele, which was operated on, hydrocephalus and polycystic kidneys presenting as abdominal masses which were confirmed by computerized tomography. In the literature, many cases of polycystic kidneys associated with anomalies of the central nervous system (CNS) have been reported[4]. Our case was diagnosed as Meckel syndrome because two of the three defining characteristics of the syndrome were present.

Although this disorder characteristically involves the CNS, the kidneys, and skeletal system, malformations of the other organ systems have also been reported. The most common anomaly involves the kidneys, however, it is possible to detect microscopic and macroscopic cysts. Associated anomalies are horseshoe kidney, renal hypoplasia and a double-collecting system. Occipital encepholocele and microcephaly are the most common anomalies of the CNS. Hypoplasia of the cerebrum and cerebellum, hydrocephalus, absence of the olfactory lobes and

pituitary gland, defects in the hypothalmic region and anencephaly are minor findings of the CNS[2]. The most common skeletal anomaly seen in this syndrome is polydactyly. Short extremities, syndactyly, simian line and clinodactyly have also been reported[3]. Anomalies which involve the face, genital organs, heart and lungs are rarely reported[2]. Meckel syndrome is inherited as an autosomal recessive trait[5]. A high incidence has been reported in some kindreds. The histories of both the subject and his twin sister (deceased) revealed encephaloceles but an unquestionable diagnosis of this disorder could not be made because of a lack of sufficient data. However, if there is a definite diagnosis made of Meckel syndrome in one sibling, then one major and some minor features of this anomaly are adequate in order to confirm this disorder in the other sibling. Since our case was definitely diagnosed as having Meckel syndrome, his sibling was also accepted as having had this disorder. As far as we know, no instance of twins having this syndrome has been reported.

The probability of Meckel syndrome occurring in a subsequent pregnancy in a family already having a member with this anomaly is 25 percent. As in other neural tube defects, such pregnancies can be terminated following prenatal tests. Determinations of alpha-fetoprotein (AFP) levels in a mother's serum in the early stages of pregnancy and in amniotic fluid cells obtained at 14-16 weeks gestation along with measurements of acetylcholinesterase in amniotic fluid to confirm the presence of a neural tube defect to eliminate false positive elevations of AFP, can be helpful[6]. Genetic counselling should be offered to couples when prenatal diagnosis is possible, as in this case.

Summary

Meckel syndrome in twins is presented. Although several families have been reported as having this syndrome in more than one member, this is the first instance that twins having this disorder have been reported. We wish to emphasize the importance of genetic counselling in such a case in which prenatal diagnosis is possible.

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