THE MECKEL SYNDROME: A Case Report and Review of Literature*

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SUMMARY

The Meckel syndrome is a rare, autosomal recessive disease. Although the diagnostic criteria are still unclear, it has been suggested that at least 2 of the 3 abnormalities (i.e.central nervous system anomaly, polycystic kidneys and postaxial polydactyly) should be present to establish the diagnosis. This report presents the pathologic findings in a stillborn female with the Meckel syndrome from Diyarbakır.

Key Words: Meckel syndrome, encepholocele, polycystic kidney, congenital malformations.

INTRODUCTION.

The Meckel syndrome, which is diagnosed by 2 of 3 main congenital malformations such as central nervous system anomaly, polycystic kidneys, and polydactyly, is an autosomal recessive disease. This syndrome was first described by Meckel in 1822. Gruber named it "Dysencephalia Splanchocystica". Opitz and Howe proposed the name "Meckel sydrome" in 1969 (1-12). The other reported cases with the Meckel syndrome (1-3) suggest that this syndrome is not rare in Turkey. A case with the Meckel syndrome which was discovered during the postmortem examination will be shortly reported.

CASE REPORT

This 3,600 gram stillborn female Turkish infant was born to a 30-year-old gravida 8, para 7 mother after a 38 weeks of gestation. The parents were first cousins and the mother denied exposure to teratogens during the pregnancy. The father is 32 years old. The family history was negative for similar birth defects on both sides.

The congenital malformations seen during the macroscopic examination revealed encephalocele, short neck, median fusion defect of the nose, cleft lip and palate, and low-set ears. In addition to these double thymus, lung hypoplasia, malrotation of intestines, left hydronephrosis, left hydroureter, and adrenal agenesis were found at autopsy, but some microscopical remnants were found in renal capsules. Histological appearance showed cystic change of kidney of variable size. No hypophyseal tissue could be detected.

DISCUSSION

Although the diagnostic criteria for the Meckel syndrome is still unclear, it has been suggested that at least 2 of the 3 cardinal signs should be present in order to establish the diagnosis. These are central nervous system anomaly, polycystic kidneys and postaxial polydactyly. This case had 2 major signs, central nervous system anomaly and polycystic kidney. The other pathological findings of this case show similarities to those reported in literature as well as differences.

Facial abnormalities were serious in this case as shown in figure 1. No other case report with similar nose fusion defect could be found.

The brain abnormalities in the Meckel syndrome, apart from the occipital encephalocele, are varying degrees of arrhinencephaly, with failure of lateralization of the forebrain (5). There may be absent olfactory bulbs and optic nerves, holotelencephaly, microgyria, basal ganglial, and brain stem malformations. The central nervous system anomaly as occipital meningoencephalocele may be associated with agenesis or hypoplasia of the cerebellum and microcephaly (6). Encephalocele can be found 4/5 of the cases.

In the present case, optic nerves were intact, but only very small portion of nervous tissue was found in encephalocele while cystic changes contining serohemmorrhagic liquid were noted. Hypothalamic as well as middle craniofacial malformations occasionally are associated with the absence of the pituitary gland which in turn may cause hypoplasia of the adrenals, and defective gonadal development. Agenesis of hypophysis was also found in our cases.

Rapola and Salonen (7,8) suggest that the adrenal aplasia was due to their abnormal position and form. Likewise, although no adrenal glad was obtained at autopsy, the adrenals were found during microscopical examination.

The thyroid gland was larger than normal contrary to other cases reported (9).

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Although postaxial hexadactyly was reported with the frequency from 74 % to 95 % in the Meckel syndrome with or without club feet joint abnormalities (1-4,7-11), in this case no abnormality of extremities was found.

Cystic changes in renal parenchyma were present in every case at different degrees. Polycystic kidneys are sometimes smaller than normal kidneys and have reduced amount of renal parenchyma with numerous small cysts, but more commonly severse polycystic involvement which may at times produce huge polycystic kidneys can be a birth impediment. The histopathological examination of this case revealed cystic change (Figs. 2,3).

Hepatic fibrosis and bile duct proliferation are often reported as additional anomalies (2,6,7,11) in patients with the Meckel syndrome. The liver of this case showed no abnormalities grossly or at microscopical examination.

Visceral anomalies such as accessory spleens, malrotation of the intestines and anal atresia have been reported (2,7,8). Only malrotation of the intestines was found in this case.

Cardiovascular and pulmonary defect may also be present (2,7,8,11). This case also showed lung hypoplasia probably due to oligohydromnios.

Various abnormalities of the genitalia including ambiguous genitalia and hypoplasia or aplasia of the uterus, vagina and penis can occur in these patients (2,7,8,11). This patient had normal genital organs.

It appears that there is a wide phenotypic expression of the mutant recessive gene. Both sexes are equally affected (6,12).

This case was stillborn. All infants in the literature were either sitillborn or died very shortly after birth except the two sisters reported by Lowry et al (13). One of these sisters lived as long as 28 months.

Atypical cases may be difficult to diagnose. Based on this experience we would be very cautious in diagnosing the Meckel syndrome in patients without minimum 2 cardinal signs. The stillborn had facial features different from the cases reported before, and had double thymus which could not been found reported previously.

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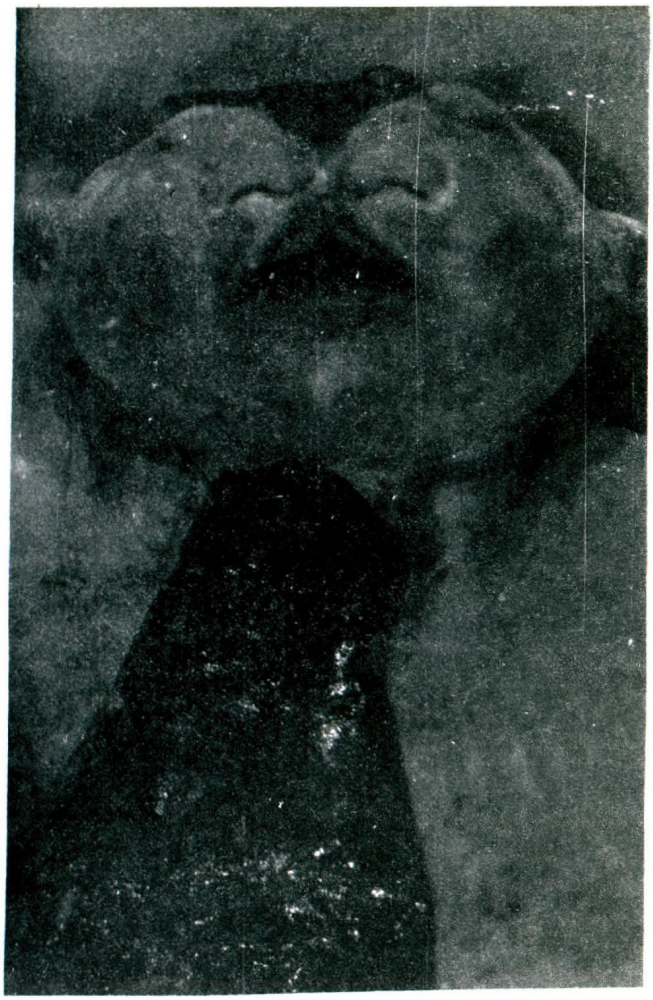

Figure 1

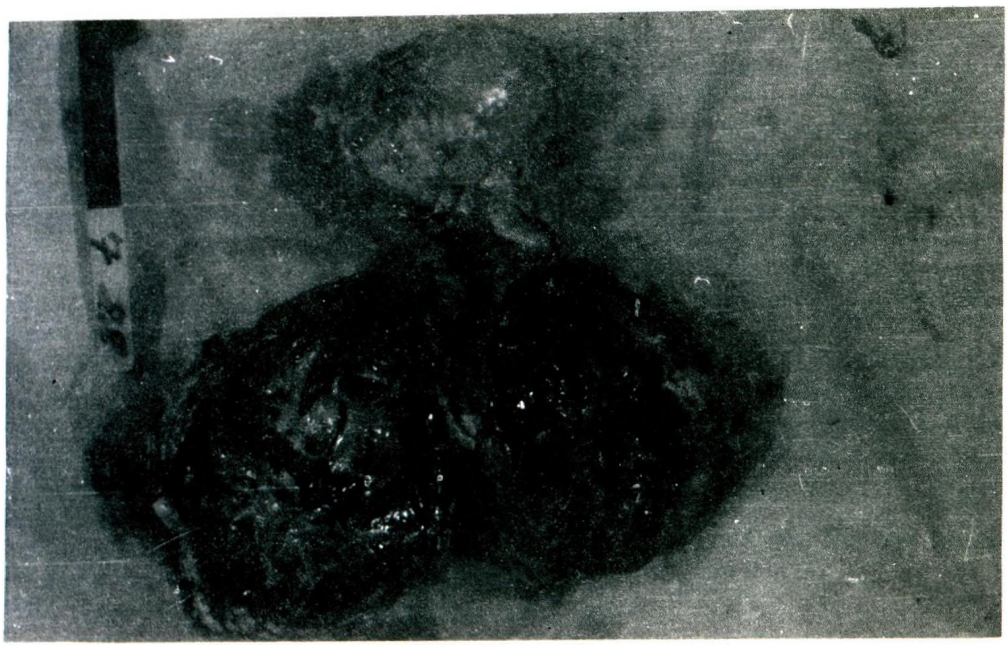

Figure 2

Figure 3

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