The Turkish Journal of Pediatrics
1974 / Vol. 16 / No. 1 / pp. 37 – 41

A Case of Omphalocele with Absence of the Thumb and Syndactily*

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Omphalocele is a protrusion of the intestines through a large defect in the abdominal wall around the umblicus.[1] The incidence of this rare and serious congential malformation is estimated to be 1 in 3200.[2] and 1 in 3800.[3] It has also been reported that omphalocele is sometimes associated with various other system malformations[2, 4] such as anomalies of the cranium, ribs and of the extremities.[5] Although a case of omphaloce le associated with phocomelia is reported by Kajii et al in 1964.[6] according to the best of our knowledge its association with absence of the thumb and syndactly has not been reported previously.

A peculiar distortion of dermal ridge arrangements has been described in cases with absence of the thumb.[7–9] The most constant findings were the transverse ridge arrangements in the proximal palm and the absence of axial triradius (t). Our case showed similiar dermatoglyphic findings.

The purpose of this communication is to report a case of omphalocele associated with thumb agenesis and its dermatoglyphic findings.

Case Report

K. B., a two-hour old male infant was admitted to Hacettepe Children's Hospital for the repair of his umblical defect. The history revealed that he was the first child born to unrelated parents. There was no one,

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on either side of the family with similar deformities. The pregnancy and labor was normal. The mother denied exposure to any known mutagenic agent.

Physical examination at birth: his weight was 3,400 g. and height 50 cm. There was a large omphalocele of 7 cm in diameter associated with agenesis of the thumb and syndactly between the second and the third as well as the fourth and fifth fingers on the right hand (Figure1). The nails of the syndactylous fingers were also hypoplastic. The vertebral X-ray and the karyotype was normal (46, XY).

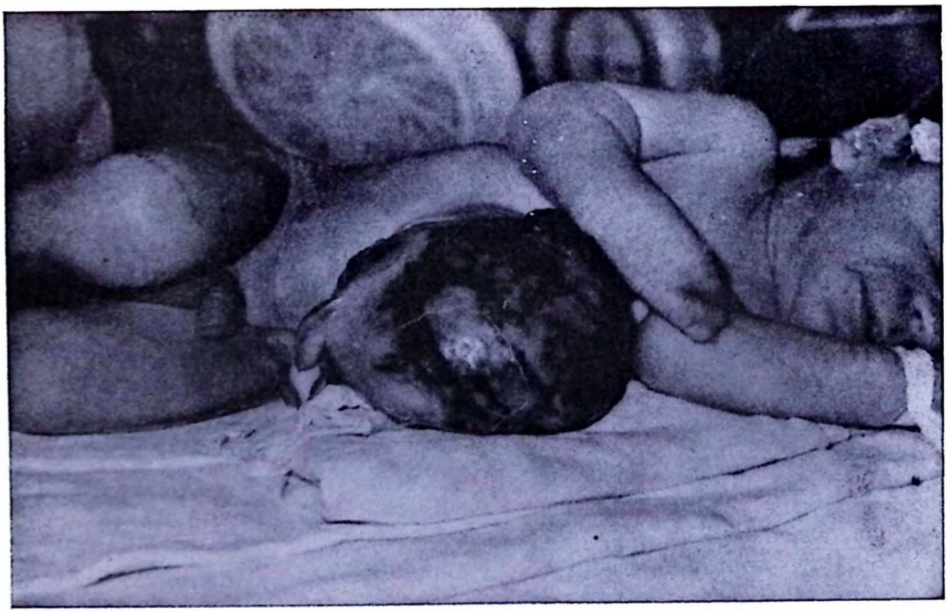

Figure 1
The appearance of the patient.

Dermatoglyphic studies showed that the aligment of the ridges on the volar surface of the left hand was normal. The only characteristic findings were intermediately, distally displaced triradii (t', t') and a high pattern intensity. However, on the right hand (which had no thumb) the ridges ran transversely in the proximal palm and there was no axial triradius. Three digital triradii typical of syndactyly were located at the base of the fused second and third, and fourth and fifth fingers. The proximal radiants of these triradii were terminated on the radial border. There were radial loops on the fourth and fifth fingertips (Figure 2). The dermatoglyphics of the parents were normal.

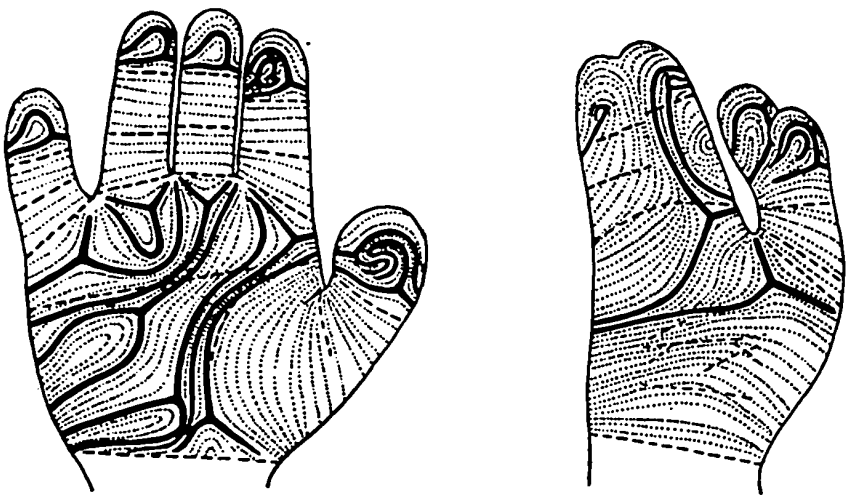

Figure 2
The dermatoglyphic of the patient.

The repair of omphalocele was done under general anesthesia. The sac contained liver as well as the bowel. Since the liver could not be replaced into the abdomen, the repair of omphalocele was done only by the approximation of the skin, instead of a primary closure.

Discussion

There is physiological herniation around the umblicus in a 6 and 11 week old embryo. But the failure of these hernia contents to re-enter the abdominal cavity in a 10 week old embryo results in omphalocele.[1] However, Kermauner believes that this malformation should occur much earlier than 10 weeks, perhaps before the begining of the third week.[5]

Although the etiology of omphalocele is still unknown, there is evidence suggesting the role of maternal infections such as hepatitis and syphilis.[10] Furthermore, omphalocele can be produced experimentally by low oxygen pressure on the ninth day of gestation in mice.[11] Similar malformations could be induced by folic acid deficiency in rats.[12] Toxic doses of salicylates have also been responsible for the omphalocele.[13]

The pecularity of our case is absence of the thumb together with omphalocele. The development of the thumb takes place between the sixth and eight weeks of the fetal life.[14] Fetal volar pads appear during the sixth and seventh weeks and begin to regress during the fourth month of gestation. The various dermatoglyphic configurations are the result

of incomplete regression of the pads before the begining of the ridge differentiation.[15] The ridges are differentiated during the third and fourth months. If, growth disturbances occurs in the foetus at this critical time, the ridge arrangements will be distorted.[15] On the other hand, Pfeiffer and Schulte Zu Berge reported the dermatoglyphics in thalidomide embryopathy with radial dysplasia. They pointed out the possible relation between aplasia of thumb and absence of the axial triradius (t) the transverse alignment of the ridges in the proximal palm.[7]

Adams noticed the same dermatoglyphic findings in a case which had absence of the thumb, ring D chromosome and multiple congenital malformations.[8] He also suggested the relation between thenar pads and distorted ridges in the proximal palm. Finally, Holt pointed out that all the cases with aplasia of the thumb, whether the other digits are present or not, show the same dermatoglyphic findings, such as the absence of axial triradius and transverse ridges.[9]

In conclusion, our case showed similar dermatoglyphic findings which were reported previously in cases with absence of the thumb. However in this patient, interestingly, absence of the thumb was associated with omphalocele. This combination might be a result of an arrest at a certain embryological stage, which was responsible for the development of both anomalies. On the other hand, it may be that these finding are merely coincidental.

Summary

A case of omphalocele associated with thumb agenesis and syndactly is presented. This combination has not been recorded previously. The patient showed characteristic dermatoglyphic findings which are described in patients with absence of the thumb. These are the transverse ridge arrangements in the proximal palm and the absence of axial triradius (t).

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