A new case of omphalocele with absence of thumb

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Omphalocele, or exomphalos, is a rare defect of the periumbilical abdominal wall in which the coelomic cavity is covered with peritoneum only, resulting in congenital evagination of abdominal contents at the base of the umbilical cord. The etiology is still unknown. In this paper a new case of omphalocele with absence of the right thumb in a five-year-old boy is reported. A common etiology and a new terminology for this combination are suggested.

Key words: omphalocele, omphalocele-radial ray defect complex (ORRD), radial ray defect, absence of thumb.

Omphalocele is a congenital defect of the abdominal wall1. The incidence is estimated to be around 1/3,800 births2. Associated malformations are not rare, especially lower chest defects, ectopia cordis and cloacal extrophy3,4.

We for the first time presented a two-hour-old male patient who had omphalocele and absence of thumb in 19745. After this observation, a case of omphalocele with partial trisomy 1q syndrome was reported by Chen at al.6 in 1979. Interestingly, this case had also bifid thumb. Later, Gershoni-Brauch et al.7 presented a case of omphalocele with absence of the left thumb and other associated anomalies in 1990. The last observation was made by Lin et al.8 in 1998 as a case of omphalocele with absent radial ray (ORR) with diploid-triploid mixoploidy. Our new case and the other cases previously reported by us and other authors suggest that the association of the omphalocele and radial ray dysplasia may not be coincidental. They may share a common etiology. A more fitting terminology for this association should be omphalocele-radial ray defects (ORRD) complex. Further cytogenetic and molecular studies of similar cases will clarify the etiology.

Case Report
A five-year-old boy presented with absence of his right thumb. This is the child of unrelated parents: the first two pregnancies resulted in spontaneous abortions. The mother was 50 years old and was treated with salicylate during the first trimester because of spontaneous abortion risk. The mother denied exposure to any other known mutagenic agents. The labor was uneventful. His birth weight was 3,900 g. The omphalocele, absence of a right thumb and inguinal hernia were noted at birth. The omphalocele was repaired at 10 months of age (Fig. 1), and inguinal hernia at two years.

His right upper extremity was relatively smaller, and his right thumb was absent (Fig. 2a, 2b). Radial styloid was palpable. Scaphoid, trapezium and first ray were not present. His fingers were normal in shape. Their range of motion and sensation were normal. Pro-supination of the forearm and elbow motion were normal. His physical examination was otherwise normal. Vertebral X-ray investigation was normal. Abdominal ultrasonographic examination and blood smear (white blood counts, Hb levels) revealed no abnormality. Chromosomal analysis of the patient and of his parents was normal.
Discussion

The etiology of omphalocele is still unknown. Folic acid deficiency, toxic doses of salicylates, and low oxygen pressure on the ninth day of gestation may have a role. Approximately half of the cases with omphalocele have associated malformations, most commonly involving cardiovascular, musculoskeletal, genitourinary, and central nervous system. The development of the thumb takes place between the sixth and eighth weeks of fetal life, while failure of re-entering of the abdominal cavity earlier than the twelfth week results in omphalocele. Unusual syndromes with omphalocele and limb malformations have also been reported.

The calculated possibility of the concurrent existence of omphalocele and radial-sided dysplasia of the upper extremity is about 1/100,000,000. Assuming incidences of 0.5 per 10,000 for radial dysplasia and 2.4 per 10,000 for omphalocele, there are only a few cases with omphalocele and radial ray malformations reported in the literature, demonstrating its low incidence. We previously reported a case of omphalocele with the absence of thumb and syndactyly. That report was followed by similar observations of an association of omphalocele with radial ray malformations.

Findings in our case and in the reported cases of omphalocele and radial-sided defects are summarized in Table I. In these cases, absence

| Table I. Published Cases of Omphalocele with Radial Ray Defects |
|-----------------|-----------------|-----------------|-----------------|-----------------|
| Birth weight (g) | 3,400          | 3,000           | 3,900          | 184 (22 wk)    |
| Sex             | Male            | Male            | Male           | Male            |
| Thumb           | R. absent       | R. bifid thumb  | L. absent, R.  | R. absent       |
| Fingers         | R2-3           | Bilateral flexion | L 2-3 syndactyly | R 4-5 syndactyly |
| Radius          | Syndactyly     | Contracture     | Contraction    | R and L radio-ulnar synostosis |
| Ulna            | —              | —               | As above       | R mildly hypoplastic |
| Humerus         | —              | —               | —              | T10 or T11 hypoplastic |
| Lower limbs     | —              | —               | —              | Normal          |
| Omphalocele     | 7 cm           | —               | Normal         | Normal          |
| Karyotype       | Normal          | 46, XY der(21) T (t(21) (g25-q27) | Normal          | 32% triploidy by flow |
| Follow-up       | Omphalocele     | Died 6 days after surgery | Died 6 days after surgery | Omphalocele repaired, healthy |
|                 | repaired        |                  | Surgery        |                  |

L: left, R: right; (—) indicates that no information was reported.
of thumb, bifid thumb, triphalangeal thumb and absence of radial ray were some of the associated malformations. All of these combinations are not possibly coincidental, and we strongly suggest that omphalocele and radial ray defects might share a common etiology. Further cytogenetic and molecular studies of similar cases will clarify the etiology.

A more fitting terminology of the association should be used such as ORRD (omphalocele-radial ray defects) complex.

REFERENCES