A new case of omphalocele with absence of thumb

Sevim Balcı[1], Gürsel Leblebicioğlu[2], İnci Yiğitkanlı[1]

Departments of [1]Pediatrics, and [2]Orthopedics and Traumatology, Hacettepe University Faculty of Medicine, Ankara, Turkey

SUMMARY: Balcı S, Leblebicioğlu G, Yiğitkanlı I. A new case of omphalocele with absence of thumb. Turk J Pediatr 2005; 47: 95-97.

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 eventration 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 wall[1]. The incidence is estimated to be around 1/3,800 births[2]. Associated malformations are not rare, especially lower chest defects, ectopia cordis and cloacal exstrophy[3,4].

We for the first time presented a two-hour-old male patient who had omphalocele and absence of thumb in 1974[5]. After this observation, a case of comphalocele 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 terminoldgy 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.

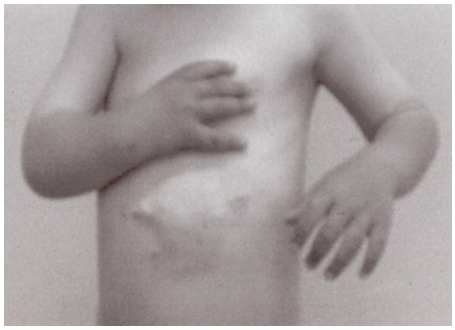
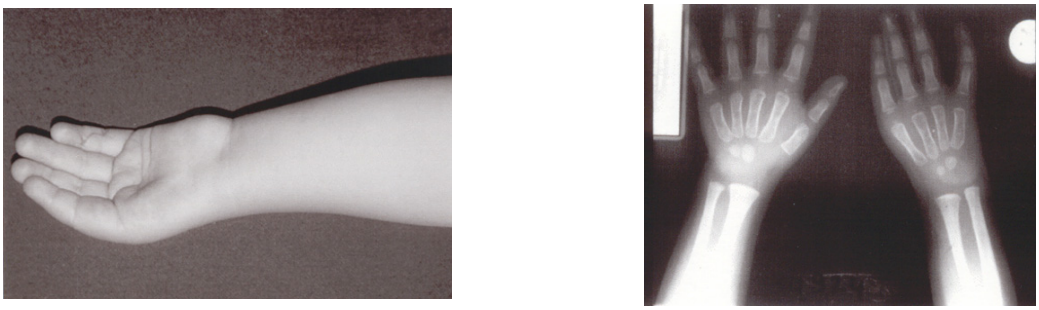


Fig. 1. Absence of the right thumb and scar from omphalocele repair.

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.



(a) (b)

Fig. 2. a) X-ray and b) appearance of the right hand showing absence of the right thumb.

Discussion

The etiology of omphalocele is still unknown. Folic acid deficiency[9], toxic doses of salicylates[10] and low oxygen pressure o the ninth day of gestation[11] may have a role. Approximately half of the cases with omphalocele have associated malformations, most commonly involving cardiovascular, musculoskeletal, genitourinary, and central nervous system[12]. The development of the thumb takes place between the sixth and eight weeks of fetal life[13], while failure of re-entering of the abdominal cavity earlier than the twelfth week results in omphalocele[2]. Unusual syndromes with ompalocele and limb malformations have also been reported[14].

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[12,15]. 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[5]. That report was followed by similar observations of an association of omphalocele with radial ray malformations[6-8,15-20].

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

Table I. Published Cases of Omphalocele with Radial Ray Defects

	Balcı et al. (1974)[5]	Chen et al. (1979)[6]	Gershoni-Brauch et al. (1990)[7]	Lin et al. (1998)[8]	Balcı et al. (2003)
Birth weight (g)	3,400	—	3,000	184 (22 wk)	3,900
Sex	Male	Male	Male	Male	Male
Thumb	R. absent	R. bifid thumb	L. absent, R. triphalangeal	Normal	R. absent
Fingers	R2-3 Syndactyly	Bilateral flexion Contracture	—	R 4-5 syndactyly, L 2-3 syndactyly	—
Radius	—	—	R and L radio-ulnar synostosis	R and L absent	—
Ulna	—	—	As above	R midly hypoplastic	—
Humerus	—	—	—	T10 or T11 hypoplastic	—
Lower limbs	—	—	Normal	Normal	Normal
Omphalocele	7 cm	—	Contained liver and intestines	Contained duodenum liver, jejunum, ileum	—
Other	—	Low set ears; presacral dimple; simian creases; micropenis; overriding toes; micrognathia cryptorchidism	R diaphragmatic hernia: midline liver with large cyst	None detected	Bilateral inguinal hernia
Karyotype	Normal	46, XY der(21) T (t:21) (q25-q27)	Normal	32% triploidy by flow (no chromosomal analysis was reported)	Normal
Follow-up	Omphalocele repaired	Died 6 days after surgery	Died 6 days after Surgery	Stillborn	Omphalocele repaired, healthy

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[5-8]. 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

1. Grosfeld JL, Weber TR. Congenital abdominal wall defects: gastrochisis and omphalocele. Curr Probl Surg 1982; 19: 157-213.
2. Soper RT, Green EW. Omphalocele. Surg Gynecol Obst 1961; 113: 501-508.
3. Smith WR, Leix F. Omphalocele. Am J Surg 1966; 3: 450-456.
4. Kirmauner D. Die Missbildungen des Rumpfes. In Schwalbe E (ed). Die Morphologie der Missbildungen des Menschen und der Tierre, Vol. 3. Jena: Gustav Fisher; 1909: 1-41.
5. Balcı S, Atasu M, Hiçsönmez A. A case of comphalocele with absence of the thumb and syndactyly. Turk J Pediatr 1974; 16: 37-41.
6. Chen H, Gershank JJ, Mailhes JB, Sanusi ID. Omphalocele and partial trisomy Iq syndrome. Hum Genet 1979; 53: 1-4.
7. Gershoni-Brauch R, Machoul I, Weiss Y, Blazer S. Unknown syndrome: radial ray defects, omphalocele, diaphragmatic hernia, and hepatic cyst. J Med Genet 1990; 27: 403-404.
8. LinHJ, Schaber B, Hashimoto CH, Barajas L, Beall MH, Lachman RS. Omphalocele with absent radial ray (ORR): a case with diploid-triploid mixoploidy. Am J Med Genet 1998; 75: 235-239.
9. Nelson MM, Wright HV, Aslin CW, Evans HM. Multiple congenital abnormalities resulting from transitory deficiency of pteroylglutamic acid during gestation in the rat. J Nutr 1955; 51: 349-369.
10. Warkany J, Takacas E. Experimental production of congenital malformations in rats by salicylate poisoning. Am J Pathol 1959; 35: 315-325.
11. Ingalls TH, Avis FR, Curley FR, Temin HM. Genetic determinants of hypoxia-induced congenital anomalies. J Hered 1953; 44: 185-193.
12. Baird PA, MacDonald EC. An epidemiologic study of congenital malformations of the anterior abdominal wall in more than half a million consecutive live births. Am J Hum Genet 1981; 33: 470-478.
13. Hamiton WJ, Boyd JD, Mossman HM. Human Embryology. Prenatal Development Form and Function. Cambridge: Heffer; 1959: 135-139.
14. Gardner RJ, Marrison PS, Faigan LA, Kennedy JC, Fitzgerald PH. Syndrome of craniofacial dysostosis, limb malformation and omphalocele. Am J Med Genet 1990; 36: 133-136.
15. Froster UG, Baird PA. Limb reduction defects in over one million consecutive live births. Teratology 1989; 39: 127-136.
16. Robinson LK, Baesl TJ, Jones KL. Omphalocele-limb abnormality sequence. Proc Greenwood Genetic Center 1987; 27: 403-404.
17. Hall BD. OURA: a new association of omphalocele and unilateral radial aplasia. Am J Hum Genet 1990; 47: A103.
18. Mastroiacovo P, Kallén B, Knudsen LB, et al. Absence of limbs and gross body wall defects: an epidemiological study of related rare malformation conditions. Teratology 1992; 46: 455-464.
19. Calzolari E, Volpato S, Bianchih F, et al. Omphalocele and gastrochisis: a collaborative study of five Italian congenital malformation registries. Teratology 1993; 47: 47-55.
20. Evans JA, Vitez M, Czeizel A. Congenital abnormalities associated with limb deficiency defects: a population study based on cases from the Hungarian Congenital Malformation Registry (1975-1984). Am J Med Genet 1994; 49: 52-66.