Concomitant Omphalocele and Anencephaly Associated with Trisomy 18 and Arthrogryposis Diagnosed in Early Pregnancy

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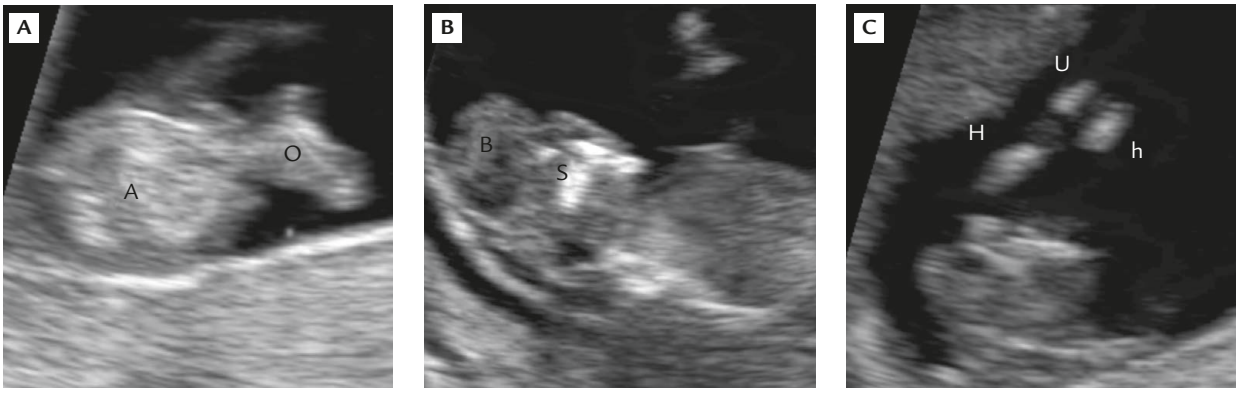

Figure 1. Prenatal ultrasound at 12 weeks' gestation shows: (A) omphalocele; (B) anencephaly; and (C) arthrogryposis of the upper limb. A = abdomen; O = omphalocele; B = brain; S = skull; h = hand; H = humerus; U = ulna.

A 41-year-old, gravida 4, para 3, woman was referred for genetic counseling because of an advanced maternal age. The woman had delivered a 20-week-gestation fetus with Down syndrome 6 years previously. Prenatal ultrasound examination at 12 weeks' gestation revealed a singleton fetus with omphalocele, anencephaly, arthrogryposis of the left hand, atrial and ventricular septal defects, and truncus arteriosus (Figure 1). The parents elected to terminate the pregnancy. A 12-g female fetus with anencephaly, omphalocele containing the intestines, and arthrogryposis of the left wrist was delivered (Figure 2). Postnatal cytogenetic analysis of the fetal tissues revealed a karyotype of 47,XX, +18. Polymorphic DNA marker analysis revealed a maternal meiosis I non-disjunction error.

We have presented the early prenatal sonographic demonstration of concomitant abdominal wall defect and neural tube defect (NTD) associated with trisomy 18 and arthrogryposis. When the diagnosis of omphalocele is made in early pregnancy, the percentage of anencephaly can increase from 61.1–66.7%, and trisomy 18 is the most common chromosomal abnormality associated with omphalocele [1]. In a study of 18 fetal omphaloceles diagnosed at 11 to 14 gestational weeks, Snijders et al [2] found that 12 cases (66.7%) had chromosomal abnormalities, of which 10 had trisomy 18. In a study of 18 fetal omphaloceles detected at 12 to 16 gestational weeks, Blazer et al [3] found that 11 cases (61.1%) had chromosomal abnormalities, of which five had trisomy 18. Chromosomal abnormalities have been reported in 2.5–10.26% of fetal and newborn patients with common NTDs, and chromosomal abnormalities occur in 0.66–5.56% of anencephaly [4]. Concurrence of abdominal wall defect and NTD associated with trisomy 18 has occasionally been reported. Moore et al [5] reported craniorachischisis, omphalocele, and bilateral cleft lip and palate in a fetus with trisomy 18. van Maldergem et al [6] reported craniorachischisis, omphalocele, bilateral radial agenesis, and distal arthrogryposis in a fetus with trisomy 18. Grangé et al [7] reported occipital encephalocele, craniorachischisis, and

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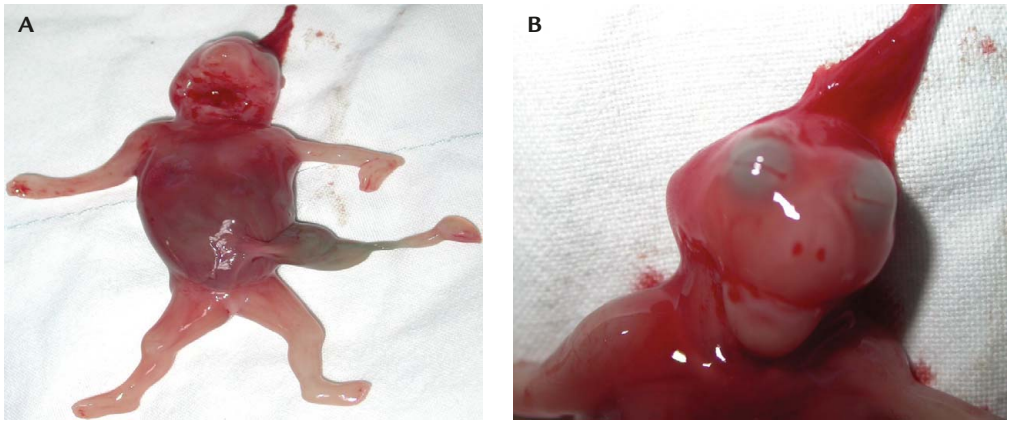

Figure 2. A 12-week-gestation fetus with omphalocele, anencephaly, arthrogryposis of the left wrist, and trisomy 18.

cystic hygroma in a fetus with trisomy 18. Donaldson et al [8] reported thoraco-abdominoschisis and craniorachischisis in a fetus with trisomy 18. Omphalocele can be associated with single gene disorders, NTDs, diaphragmatic defects, fetal valproate syndrome, and syndromes of unknown etiology [9]. Calzolari et al [10] proposed that omphalocele and NTDs are related congenital anomalies by the findings of a tendency for omphalocele to be associated with anencephaly and/or spinal bifida. Prenatal diagnosis of concomitant abdominal wall defect and NTD should include a differential diagnosis of amniotic band syndrome and limb–body wall complex with craniofacial defect [11,12]. As shown in this presentation, prenatally detected arthrogryposis may provide a clue to trisomy 18, since about one-fourth of the cases with fetal trisomy 18 have arthrogryposis of the wrist and/or ankle [13].

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