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Letter to the Editor

Recurrent omphalocele with partial trisomy 3q and partial monosomy 11q

To the Editor:

Omphalocele is not an uncommon malformation; however, it usually occurs sporadically with less than a 1% recurrence risk (Winter & Baraitser 1991). We present recurrence of omphalocele in siblings with partial trisomy 3q and partial monosomy 11q as the unbalanced product of a t(3;11)(q21;q23)mat.

This was the fourth pregnancy of an unrelated couple. The mother had one healthy 5-year-old daughter, one miscarriage and one stillbirth with omphalocele. The mother had a balanced reciprocal translocation: 46,XX,t(3;11)(q21;q23) (Fig. 1). The father had a 46,XY karyotype. Their previous stillbirth demonstrated an omphalocele in combination with an abnormal chromosome complement of 46,XX,der(11),t(3;11)(q21;q23)mat. In this pregnancy, an omphalocele containing the liver, short limbs and intrauterine growth retardation were observed at 18 weeks' gestation. Amniocentesis further revealed an abnormal karyotype with partial trisomy 3q and partial monosomy 11q as the unbalanced product of a 46,XX,t(3;11)(q21;q23)mat (Fig. 1). The pregnancy was terminated at 22 gestational weeks. Neonatal external abnormalities (Fig. 2 and 3) included a prominent keel-shaped forehead, hypertelorism, anteverted nostrils, high-arched palate, a long philtrum, a short broad nose with a wide, depressed nasal bridge, epicanthic folds, a carp-shaped mouth with thin vermilion borders and down-turned corners, micrognathia, low-set ears, a short neck, widely spaced nipples, and a 6 × 6 cm omphalocele. Internal examination at autopsy showed an omphalocele containing liver and intestines. The other visceral organs, uterus, tubes and ovaries were normal. There were no digital abnormalities.

In the presence of duplication of 3q21→qter and deletion of 11q23→qter, the recurrence of a ventral midline defect in this case is significant. Omphalocele has not been listed as one of the clinical or associated findings in either distal trisomy 3q or partial monosomy 11q syndromes in most recent reports (Donnenfeld et al. 1990, Fryns 1990,

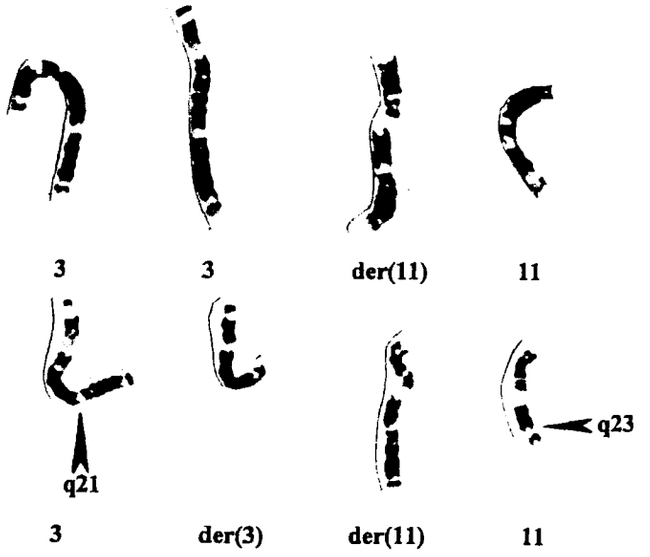

Fig. 1. Chromosome preparation showing (upper) the chromosomes 3, 11 and der(11) in the proband and (lower) the balanced reciprocal translocation identified in the mother (arrows indicate the breakpoints).

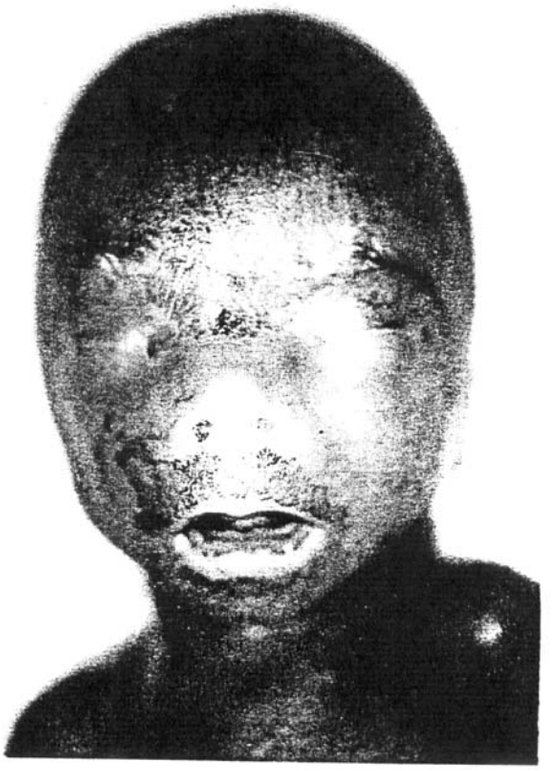

Fig. 2. Anterior view of craniofacial dysmorphism in the proband.

Fig. 3. Lateral view of omphalocele with an extracorporeal liver.

Schwarz et al. 1992, Aqua et al. 1995, Lewanda et al. 1995). However, Schinzel (1983) previously pointed out that dup(3)(q25→qter) was associated with omphalocele. Omphalocele had been reported in dup(3q)/del(3p) syndrome (Allderdice et al. 1975, Fineman et al. 1978, Steinbach et al. 1981). We previously reported the first case of omphalocele associated with partial duplication of 3q and distal deletion of 11q (Chen et al. 1996). This is the second affected female fetus with omphalocele and the same chromosomal duplication/deficiency in this family. This aberration indicates that 3q duplication/11q deficiency is likely to be associated with ventral midline defects.

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