De novo inverted duplication of chromosome 7q

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Abstract

We report a fetus with a de novo inverted duplication of the long arm of chromosome 7, karyotype 46,XX, inv dup(7) (pter→q36.1::q36.1→q22::q36.1→qter).

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Duplications are uncommon events; a search of published reports showed only 40 previous reports of any duplication involving the long arm of chromosome 7. Two of these reports were of de novo duplications. In four the duplication was the sole chromosomal abnormality and in only one case were the breakpoints similar to the case presented here.

Case report

This fetus was terminated at 18 weeks of pregnancy because of hydrocephalus. The mother was a 26 year old primigravida. The female fetus weighed 330 g, with a crown-rump length of 20 cm, crown-heel length of 29 cm, and foot length of 24 mm, consistent with gestation, and a head circumference of 19 cm, consistent with the prenatal diagnosis. The occiput was rather prominent. There were downward slanting palpebral fissures with apparent hypertelorism and a broad nasal root with an anteverted nose (fig 1). The palate and neural tube were intact. The pelvis appeared relatively small with an anteriorly placed anus. There was bilateral talipes equinovarus. The hands appeared normal. The placenta was unremarkable with three vessels in the cord. Internal examination showed a brain with normal formation of the cerebral hemispheres, falx, and tentorium cerebelli. The description macrocephaly is thus more appropriate than hydrocephalus. The other internal organs also appeared entirely normal with the exception of the uterus which was unicornuate with absence of the right fallopian tube, but both ovaries appeared normal. Karyotyping of cultured skin fibroblasts showed extra chromosome material attached to the long arm of one chromosome 7, which appeared to be the result of an inverted duplication of 7q22→q36.1, the karyotype being 46,XX,inv dup(7) (pter→q36.1::q36.1→q22::q36.1→qter) (fig 2). Both parental karyotypes were normal. No cell line is available from this patient.

Discussion

Forty-one cases of duplication of chromosome 7q have been reported including the present case. Six cases have breakpoints in the region 7q21→7qter, but only one has the same breakpoints (7q22→7q36.1) as the case reported here.¹ There are a number of similarities in the

Figure 1 The fetus showing macrocephaly, downward slanting palpebral fissures, and anteverted nose.

Figure 2 Partial karyotype showing the inverted duplication of chromosome 7 on the left and the normal 7 on the right. Breakpoints are indicated by arrows on the ideogram of chromosome 7.

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phenotypes of the two cases, such as macrocephaly, prominent occiput, hypertelorism, downward slanting palpebral fissures, and a broad flat nasal root. Macrocephaly and prominent occiput were also reported in the other cases with breakpoints between 7q21 and 7qter. In the present case duplication of part of 7q was the only chromosome anomaly and therefore the traits reported by us are in all probability the result of this anomaly. The only features that were not also reported by Forabosco *et al*¹ are bilateral talipes equinovarus and unicornuate uterus with absent right fallopian tube; however, no necropsy was performed on their case. Dysmorphic features reported by Forabosco *et al*¹ but not by us, such

as wide fontanelles, frontal bossing, macroglossia, kyphoscoliosis, and skeletal anomalies, are probably the result of deletion of chromosome 1 (q22→qter) also present in their case. These features are unlikely to be the result of the duplication as they were not reported in other cases.

This case is important because it helps define the phenotypic effects of duplication of part of the long arm of chromosome 7 and lends support to macrocephaly as an unusual feature present in duplications of 7q21→qter.

1 Forabosco A, Baroncini A, Dalpra L, *et al*. The phenotype of partial dup(7q) reconsidered: a report of five new cases. *Clin Genet* 1988; 34:48–59.