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Short report

Del(3)(p25.3) without phenotypic effect

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Abstract

A terminal deletion of chromosome 3 at p25.3 was observed during prenatal diagnosis. A similar deletion is also present in the phenotypically normal mother. The deletion was confirmed by FISH. The breakpoint is distal to the region responsible for the 3p- syndrome. A normal baby girl was born with no apparant phenotypic abnormalities.

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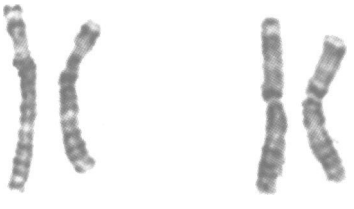

Figure 1 GTG banded partial karyotype of the fetus (left) and the mother (right). In each case the deleted chromosome 3 is shown on the right.

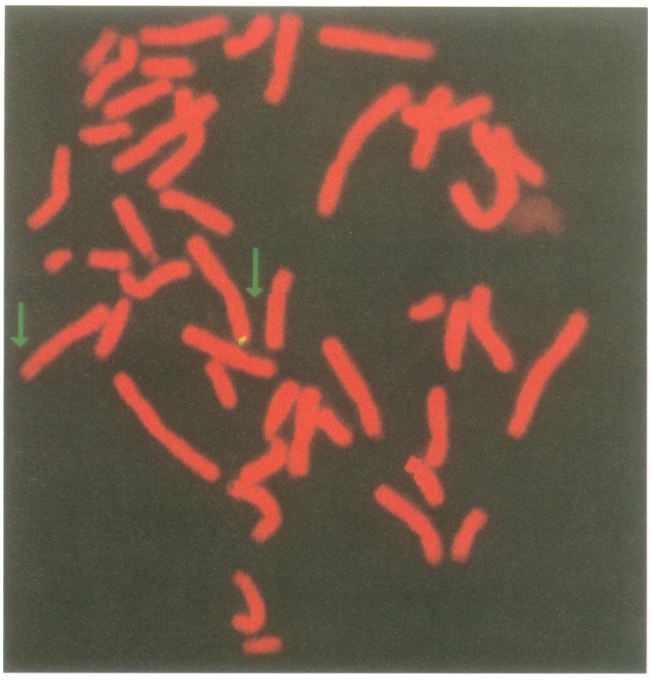

Figure 2 Metaphase from the mother after FISH with ONCOR Tel 3p DNA probe. Arrows indicate chromosomes 3.

Terminal deletion of chromosome 3 from p25 to pter is a rare chromosome deletion associated with severe congenital abnormalities. Over 20 cases have been described so far. Most have growth failure, psychomotor and mental retardation, craniofacial anomalies, and postaxial polydactyly.[1 2]

A 36 year old gravida 2 para 1 woman had amniocentesis performed for advanced maternal age. Chromosome analysis showed an abnormal fetal karyotype with a terminal deletion of the short arm of chromosome 3 at p25.3. All other chromosomes were normal except for an inv(9)(p11q13). The father's phenotype was normal and he carried the inv(9). A terminal deletion of the short arm of chromosome 3 at p25.3 was also found in the phenotypically normal mother (fig 1) and again all other chromosomes appeared normal. Fluorescence in situ hybridisation with a DNA telomere probe specific for 3p26 to 3pter detecting loci D3S1442, D3S1443, and D3S1444 (ONCOR) showed hybridisation signals to only one of the mother's chromosomes 3 (fig 2). This means that she carries a deletion for this terminal band.

Since the mother was phenotypically normal with no physical or mental handicap, she was advised to continue the pregnancy. The baby was born healthy and physical examination showed no structural abnormalities. A detailed clinical examination was carried out on the mother and child and none of the characteristic features related to the 3p deletion syndrome was observed.

The cytogenetic breakpoint of the deletion associated with the 3p- syndrome has been identified as 3p25. The variability in the phenotypic spectrum might be related to differences in the extent of the deletion. Phipps *et al*[3] performed molecular genetic analysis in five cases of 3p- syndrome and found that the extent of the deletion was variable with breakpoints between RAF1 and D3S1317 in four patients and telomeric to D3S1317 in one patient. They concluded that the loss of sequences centromeric to D3S1317 is not required for expression of the characteristic 3p- syndrome phenotype. Drumheller *et al*[4] analysed the breakpoints of the deletion in three 3p- syndrome patients. They found the breakpoints to be localised distal to D3S1038 but

proximal to D3S18. The findings of these two studies are concordant.

In the present family, the breakpoint of the deletion has been identified as p25.3. Since both the child and mother have a normal phenotype, the breakpoint must be distal to the genes responsible for the 3p- syndrome. As evidenced by this family, a deletion distal to 3p25.3 has no apparent deleterious effect.

1 Asai M Ito Y, Iguchi T, Ito J, Okada N, Oishi H. Terminal deletion of the short arm of chromosome 3. *Jpn J Hum Genet* 1992;**37**:163–8.
2 Nienhaus H, Mau U, Zang KD. Infant with del(3)(p25-pter): karyotype-phenotype correlation and review of previously reported cases. *Am J Med Genet* 1992;**44**:573–5.
3 Phipps ME, Latif F, Prowse A, *et al.* Molecular genetic analysis of the 3p- syndrome. *Hum Mol Genet* 1994;**3**: 903–8.
4 Drumheller S, O'Brien S, Roberson J, Van Dyke DL, Smith DI. Characterization of the chromosome 3 breakpoints in three patients with the 3p- syndrome. *Am J Hum Genet Suppl* 1994;**55**:A102.