

Extra G positive band on the long arm of chromosome 9

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Abstract

Various heteromorphisms of the 9q heterochromatic area have been reported. In most instances, the extra G positive band is accompanied by an extra C band. We describe a family where the extra G band is totally euchromatic and does not include an extra C band. It is not clear whether these two types of variant chromosome 9 arose from a similar mechanism.

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In recent years, there have been reports of rare variant chromosome 9. Roland *et al*¹ reported two unrelated families with an inherited chromosome 9 where an extra G positive band was found adjacent to the heterochromatic region of the long arm.

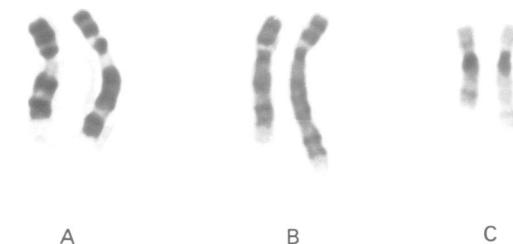
Previously Luke *et al*² described a similar chromosome in a patient with mental retardation and behavioural problems. The mother's karyotype was normal and the father was unavailable for analysis. Thus it is not possible to exclude that this chromosome 9 was inherited and the observed phenotype might not be the result of the abnormal chromosome 9.

Case report

A 37 year old primigravida had amniocentesis performed for advanced maternal age. This was her first pregnancy after a four year period of subfertility.

Chromosome analysis showed an extra G dark band on the long arm of one chromosome 9 in all cells examined. All other chromosomes were normal. This observation was further confirmed on a fetal blood specimen. The father's karyotype was normal. The anomalous chromosome 9 was also found in the phenotypically normal mother. The excess material involving 9q13-22 was C band negative (figure). No cell line is available from this patient.

Since the mother was phenotypically normal with no physical or mental handicap, she was advised to allow the pregnancy to continue. Antenatal progress was uneventful and repeated ultrasound scans showed no abnormalities. She finally gave birth to a baby boy at term. The boy was assessed to be normal by the paediatricians at birth and is at present being followed up by them.



GTG banded partial karyotype of the fetus (A) and mother (B), CBG banding (C).

Discussion

There appears to be two types of variant chromosome 9q described in published reports. The first type consists of an extra G band within the heterochromatic region. A small extra G band located within a large heterochromatic region detected in 3 to 50% of cells in various subjects was first described by Madan.³ In the case reported by Docherty and Hultén,⁴ the extra band was large and easily identified in all cells. This anomalous chromosome 9 was described in a child with trisomy 21 Down's syndrome, its phenotypically normal mother, and one of the two normal brothers. Similarly, in the two unrelated families published by Roland *et al*¹ and also the case reported by Luke *et al*,² there were two C bands in the secondary constriction in addition to the centromeric heterochromatin.

In the second type which includes our observation and the family reported by Jalal *et al*,⁵ the extra material was euchromatic and C band negative. It is surprising that the presence of a significant amount of euchromatic material should be without phenotypic effect. The mechanism involved is not clear and some authors have suggested gene amplification. For diagnostic purposes, parental studies should always be included.

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- 4 Docherty Z, Hultén MA. Extra euchromatic band in the qh region of chromosome 9. *J Med Genet* 1985;22:156-7.
- 5 Jalal SM, Kukulich MK, Garcia M, Day DW. Euchromatic 9q+ heteromorphism in a family. *Am J Med Genet* 1990;37:155-6.

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