two centromeres indicated by two distinct C bands but only one primary constriction at the proximal C band. The two C bands were separated by chromosomal material staining pale in G banding and intensely dark in R banding (fig 1). Both NORs could be observed in satellite associations (fig 2). The chromosome was therefore defined as pseudodicentric chromosome 21 (pseudic 21). The same chromosome was found in the proband's father and paternal grandmother.

Acrocentric chromosomes with a short arm morphology similar to that presented here have been reported by Balíček and Žižka.¹ These authors paid no attention to the activity of the centromeres. The suppression of additional centromeres is indicated by the presence of only one primary constriction as shown by Ing and Smith² in a dicentric (Y;13) translocation. Variants of acrocentric chromosomes are often observed in patients with congenital anomalies.¹⁻³ The occurrence of the pseudodicentric chromosome 21 in the proband and her phenotypically normal father and grandmother indicates that there is no association between the chromosomal variant and the proband's congenital anomalies.

FIG 1 Chromosomes 21 of the proband (a) and her father (b) by G (GAG), R (RHG), C (CBG) banding, and silver staining (Ag-NOR).

FIG 2 Pseudodicentric chromosome 21 in satellite associations. (a) G bands, (b) R bands.

I HANCKE AND K MILLER
Department of Human Genetics,
Medizinische Hochschule Hannover,
Hannover,
Federal Republic of Germany.

References

- Balíček P, Žižka J. Intercalar satellites of human acrocentric chromosomes as a cytological manifestation of polymorphisms in GC-rich material? *Hum Genet* 1980;54:343-7.
- Ing PS, Smith SD. Cytogenetic studies of a patient with mosaicism of isochromosome 13q and a dicentric (Y;13) translocation showing differential centromeric activity. *Clin Genet* 1983;24:194-9.
- Passarge E. Analysis of chromosomes in mitosis and evaluation of cytogenetic data. 5. Variability of the karyotype. In: Schwarzbacher HG, Wolf U, eds. *Methods in human cytogenetics*. Berlin, Heidelberg, New York: Springer, 1974:167-77.

Correspondence and requests for reprints to Dr K Miller, Department of Human Genetics, Medizinische Hochschule Hannover, PO Box 610180, D-3000 Hannover 61, Federal Republic of Germany.

Extra euchromatic band in the qh region of chromosome 9

Chromosome 9 variants with a small extra G band located within a large heterochromatic region in the long arm were first described by Madan,¹ the extra band being detected in 3 to 50% of cells in various subjects, and similar variants have since been reported by other authors. We describe here an unusual variant 9 in which the extra band is large and easily identified in all the cells examined.

The anomalous chromosome 9, first detected in a child with primary trisomy 21, is also present in the mother and in one of two phenotypically normal brothers. The cytogenetic characteristics are illustrated in the figure. The extra band exhibits medium fluorescence with Q and sequential Q/C staining. Three C bands were seen in the variant, interpreted as representing the heterochromatin of the centromere region, and the proximal and distal parts of the heterochromatin of the secondary constriction flanking the extra negatively stained band. In contrast, the C positive region in the homologue has two subunits.

The extra band gave a negative reaction with a silver staining method (Goyanes,² first step), which shows a staining affinity roughly coinciding with the loci rich in satellite III DNA. This method stains the secondary constriction heterochromatin leaving the centromeric heterochromatin unstained. In the variant, two dark segments were seen with a negatively stained band between them, indicating that the extra band is located within 9q12. This was confirmed with distamycin A/DAPI and G11 staining.

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FIGURE Variant chromosome 9 (arrowed) identified by (a) G-banding, (b) C-banding, (c) silverstaining.²
 (d) Morphological organisations of the heterochromatic region in chromosome 9 as proposed by the authors.
 (i) Common type of chromosome 9 with a G positive band (Gb1) between the pericentromeric heterochromatin and the secondary constriction heterochromatic that is, between bands q11 and q12.
 (ii) Our variant, with an extra G positive band (Gb2) located within the secondary constriction heterochromatic block (q12).

The only other reported case with such a large extra G band seen in all the cells³ was also familial and ascertained via a child with Down's syndrome but this association is most likely to be fortuitous. We may be dealing here with an extreme variant in the size of the extra G band which is not uncommon within the 9qh region but which is usually much smaller and therefore only detected in some cells.¹ We have noticed such smaller additional G bands within the 9qh region in seven out of 500 cases (1.4%) during diagnostic screening, and we find that this type of heteromorphism is even more common in chromosome 1, where it was seen in over half of the 500 cases. Here, however, the G banding pattern is the reverse of that in

chromosome 9, that is, a light G band within the dark qh region.

We believe that the normal morphological organisation of the heterochromatic region of chromosome 9 is as shown in the figure, with a dark G band (Gb1) between the pericentromeric heterochromatin and the heterochromatin of the secondary constriction. An increase in the amount of pericentromeric heterochromatin would displace band Gb1 to a more distal location giving the impression of an additional band, similar to the situation which occurs in the p arm, with 9p12 appearing as an extra band. Thus we are not certain that some of the chromosomes which we have interpreted as having an extra band are not a false impression and the same may be true of some of the published cases. In the variant presented here, however, and also in some of the published cases where G11 staining was carried out, it is clear that some additional material is present which is not constitutive heterochromatin and this is illustrated schematically in the figure.

It is of interest that all the variants described so far as having an extra G band within the 9qh have been associated with relatively large qh blocks and that the only reported de novo case involved not only the occurrence of an extra G band but also an increase in the amount of the surrounding heterochromatin.¹ It therefore seems unlikely that the extra band represents a simple insertion and more likely that this whole region has become amplified. Whatever the mechanism(s) behind the amplification, it appears to have been associated with inactivation of any functional genes located here. Alternatively, such genes may have or have had a regulatory function in the development of a common polymorphic system such as, for example, antibody formation and immunity. Any such phenotypic effect may easily escape detection.

ZOE DOCHERTY AND MAJ A HULTÉN
 Regional Cytogenetics Laboratory,
 East Birmingham Hospital,
 Bordesley Green East, Birmingham B9 5ST.

References

- Madan K. An extra band in human 9qh+ chromosomes. *Hum Genet* 1978;43:259-64.
- Goyanes VJ. Sequential staining of euchromatic and heterochromatic regions of the human Y chromosome. *J Med Genet* 1980;17:468-71.
- Berg JM, Gardner HA, Gardner RJM, et al. Dic(21:21) in a Down's syndrome child with an unusual chromosome 9 variant in the mother. *J Med Genet* 1980;17:144-8.

Correspondence and requests for reprints to Dr Zoe Docherty, Department of Cytogenetics, Infant Development Unit, Birmingham Maternity Hospital, Queen Elizabeth Medical Centre, Edgbaston, Birmingham B15 2TG.