A new stable human dicentric chromosome, tdic(4;21)(p16;q22), in a woman with first trimester abortion

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Abstract

A woman with first trimester abortion and a dicentric chromosome formed from a 4 and a 21 is described. The dicentric chromosome was stable and in the majority of cells the 21 centromere was active, while in a minority the chromosome 4 centromere was active. This shows that both centromeres were functional, but that only one functioned in any given cell. Suppression of the activity of one centromere might be the mechanism by which this dicentric chromosome achieved its stability. A dicentric formed from a chromosome 4 and a 21 has not apparently been previously reported.

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Case report

A 32 year old woman presented for genetic counselling because of two first trimester abortions. She had no history of illness or drug ingestion and the fetuses were aborted at about 8 weeks' gestation. There was no family history of miscarriage or multiple congenital anomalies. Clinical examination was normal.

Chromosome analyses were performed on peripheral lymphocytes and skin fibroblasts. In all cells examined a dicentric chromosome formed from a chromosome 4 and a 21 through an end to end translocation was observed. In the majority of cells (96%) the 21 centromere was active while in a minority (4%) the chromosome 4 centromere was active (figure). The karyotype of this patient can be described as 45,XX,-4,-21,+tdic(4;21)(p16;q22). The aberration occurred de novo as the parents were cytogenetically normal. Unfortunately, a cell line is not available from this patient.

Discussion

In rare instances dicentric chromosomes occur as constant members of a chromosome complement. With the exception of those in which the two centromeres are close together, dicentrics may achieve their stability by two mechanisms: (1) suppression of the activity of one centromere, or (2) visually undetectable deletion of one centromere.¹ Asymmetrical dicentric chromosomes are usually members of a 45 chromosome complement in which the likelihood of viability is increased by one or both of the participants being acrocentric.² In almost all dicentrics that have been reported, the active centromere was always the same in each subject. In a dicentric formed from an acrocentric and a non-acrocentric, the dominant centromere might be from either form.^{3,4} In some cases the two centromeres appear to be alternately inactivated.² The probable explanation is that an originally different centromere was inactivated in different cells, or a reversion of centromere inactivation occurred to produce both dicentric cell lines.

A stable dicentric chromosome formed from a chromosome 4 and a 21 has not apparently been reported previously. In the present case, the 21 centromere was active in the majority of cells, while in a minority the centromere of chromosome 4 was active. All dicentrics observed showed two primary constrictions and there were no deletions. This shows that both centromeres were functional, but that only one functioned in any given cell. Suppression of the activity of one centromere might be the mechanism by which the dicentric chromosome achieved its stability.

We do not know exactly how and when one centromere of a dicentric is inactivated but we assume that one centromere suppressed another just as the dicentric formed. The normal phenotype of the present case indicates intact genetic function. However, the first trimester abortions might have been the result of her chromosomal abnormality.

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Partial karyotype of the patient. The abnormal chromosome is in the middle with the normal chromosome 4 on the left and the normal chromosome 21 on the right of each group. (A) Giemsa staining, (B) GTG banding, (C) and (D) C-banding.

- 1 Therman E, Sarto GE, Patau K. Apparently isodicentric but functionally monocentric X chromosome in man. *Am J Hum Genet* 1974;26:83-92.
- 2 Dewald GW, Boros SJ, Conroy MM, et al. A tdc(5;15)(p13;p11) chromosome showing variation for constriction in the centromeric regions in a patient with the cri du chat syndrome. *Cytogenet Cell Genet* 1979;24:15-26.
- 3 Schwartz S, Palmer CG, Weaver DD, et al. Dicentric chromosome 13 and centromere inactivation. *Hum Genet* 1983;63:332-7.
- 4 Vianna-Morgante AM, Rosenberg C. Deletion of the centromere as a mechanism for achieving stability of a dicentric chromosome. *Cytogenet Cell Genet* 1986;42:119-22.