

Material

Pericentric Inversion in Homologues of Chromosome 9

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Introduction

Pericentric inversion in one of the chromosomes of 9 is a commonly observed structural variation in the human population and this is transmitted as a dominant character, since it is not known to result in deleterious consequences (De La Chapelle *et al.*, 1974). This anomaly is also observed in association with congenital abnormalities and reproductive loss (Boue *et al.*, 1975; Tibiletti *et al.*, 1981; Sudha and Gopinath, 1991).

An instance of pericentric inversion in both the homologues of chromosome 9, was observed in a female child with labial adhesion. In literature, five individuals have been reported with such condition, having different phenotypes (Wahrman *et al.*, 1972; De La Chapelle *et al.*, 1974; Vine *et al.*, 1976; Sakagami *et al.*, 1988).

Case history

The female child (11/2 years) was referred for chromosomal analysis due to the fusion of labia minora. The line of adhesion was firm and thick. Labia majora was well formed and urethra and clitoris were present. The ultra-sonographic report revealed the following details: uterus 2.6 × 0.7 cm; right ovary 1.2 × 0.6 cm; left ovary 1.0 × 0.6 cm; giving an impression of normal uterus and ovaries. The child was active and intelligent.

Proband was the only child of a consanguineously married couples (first cousins, once removed—Fig. 1). Instances of deaf mutes, blood cancer and fetal loss were reported in some of the family members.

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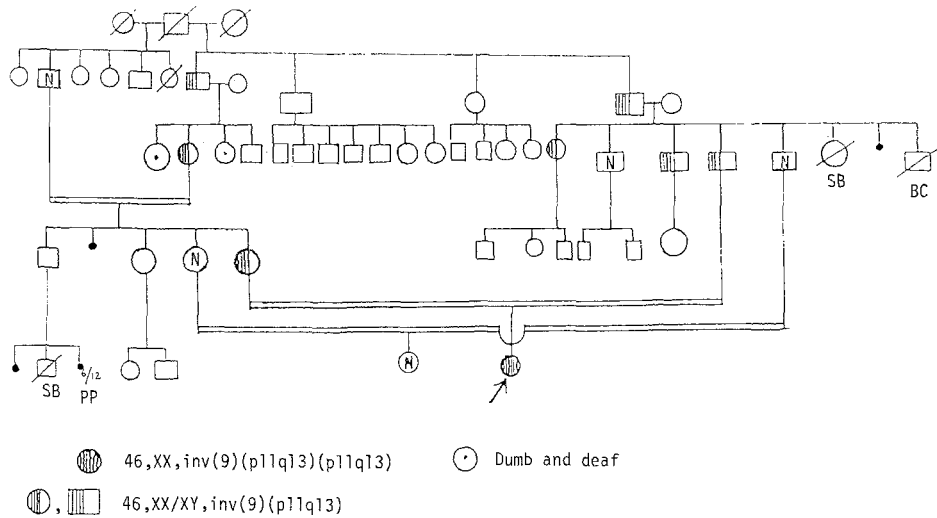


Fig. 1. Pedigree showing the transmission of pericentric inversion of chromosome 9 leading to inversion in homologues.

Cytogenetic analysis

Leucocyte cultures were established. Cytogenetic analysis of the G-banded metaphases of the proband, revealed inversion 9 in the homologues and the karyotype was 46,XX,inv(9)(p11q13),(p11q13). C-banding was carried out which confirmed the pericentric inversion. This inversion was found to be inherited from her parents, who had inv(9) in heterozygous condition. Further, cytogenetic investigations of ten other family members showed the anomaly in five of them, with no phenotypic abnormalities (Fig. 1), and also revealed its common origin.

Except in the patient described by Sakagami *et al.* (1988), the homozygous condition of inv(9) was a result of consanguineous marriages in the parents. Mental and growth retardation and other clinical abnormalities were observed by De La Chapelle *et al.* (1974) and Sakagami *et al.* (1988) when this variation was recorded in homozygous condition. However, no such phenotypic anomalies were seen in the present case and also in the other reports by Wahrman *et al.* (1972) and Vine *et al.* (1976).

It can be considered that, the inversion in chromosome 9 as a casual co-existence with different clinical abnormalities and there seemed to be no definite correlation with homozygous inv(9) condition and phenotypic anomalies. However, members of this particular family may be useful as a source of DNA analysis of chromosome 9, homozygosity.

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