Cases Observed

Balanced Reciprocal Translocation t(1;16)(q12;q11) in Two Generations of a Family

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A 7-year-old female with normal intelligence but short stature (105 cm) was found to be a carrier of a balanced reciprocal translocation 46,XX,t(1;16)(q12;q11). Family studies demonstrated carriership of her mother, who also had normal intelligence, a height of 167 cm, and up to now two abortions.

A translocation t(1;16) has been found in two previous cases (Friedrich and Hielsen 1973; Fried et al. 1977), the second one in three generations of a family associated with frequent abortions.

References

- Fried K, Perpinyal S, Rosenblatt M, Caspi E (1977) Familial balanced reciprocal translocation t(1;16)(q12;q13) ascertained because of multiple abortions in a carrier. *Hum Hered* 27:362–365
- Friedrich U, Hielsen J (1973) Chromosome studies in 5,049 consecutive newborn children. *Clin Genet* 4:333–343