Hum Genet (1981) 56:421

Human Genetics

Letters to the Editors

Trisomy 22 Mechanisms

H. H. Punnett and M. L. Kistenmacher

Genetics Laboratory, St. Christopher's Hospital for Children, 5th and Lehigh, Philadelphia, PA 19133, USA

In 1971 we described the first two cases of "proven" trisomy 22 using quinacrine fluorescence (Punnett et al. 1971). In a third patient, studied using trypsin giemsa banding, (Punnett et al. 1973) one of the chromosomes 22 appeared to be shorter. This deleted 22 was also seen in four normal members of the family. (No size difference was noted in the two original patients studied using quinacrine banding.)

It was assumed that the deleted 22 was one member of a pair of chromosomes comprising a balanced translocation. The other member was not identified. Lejeune (1972) was the first to recognise, in a sterile male, the involvement of chromosome 11 in this rearrangement. Subsequently there have been many reports of normal family members with the balanced translocation (11;22)(q23;q11), usually discovered through a propositus with an extra deleted 22. Ferguson-Smith (1978) has suggested that this may be one of the most common reciprocal rearrangements in man. The affected offspring have tertiary trisomy 22;11 resulting from 3:1 meiotic segregation in a parent, usually the mother.

Recently, in this Journal, Biederman et al. (1980) compared the expression of full trisomy 22 with tertiary 22. Our original patients 1 and 2 were included in the table as having the full trisomy. We wish to correct the record. We have restudied these patients and discovered that both propositi carry the derived 22;11. The mother of SC (Patient 1) and the father of MR (Patient 2) carry the 11;22 translocation. We suggest that other patients diagnosed as having trisomy 22 should be reevaluated, as well as their parents and siblings, in view of the reproductive risk for family members carrying the 11;22 translocation. The question of the viability of true trisomy 22 remains unanswered at this time.

Acknowledgement. Supported in part by USPHS Grants CA 19834 and RR 75.

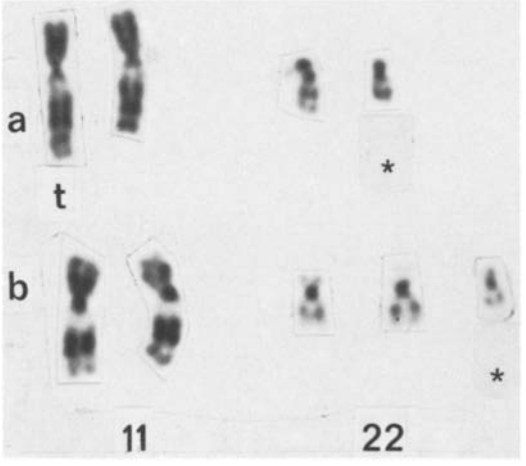

Fig. 1. Partial karyotypes, GTG banded, of SC (*b*) and his mother (*a*). *t* = 11;22 and * = 22;11

References

Biederman BM, Lin CC, Lowry RB, Somerville R (1980) Tertiary trisomy (22q11q),47,+der(22),t(11;22). Hum Genet 53: 173–177

Ferguson-Smith MA (1978) Human cytogenetics. Birth Defects 349–357

Lejeune J, Dutrillaux B, Pethore M-O, Prieur M, Couturier J, Carpentier S, Raoul O (1972) Analysis of 30 cases of translocation by the controlled heat denaturation. In: Modern aspects of cytogenetics; Constitutive heterochromatin in man. F. K. Schattauer Verlag, Stuttgart, pp 193–198

Punnett HH, Kistenmacher ML, Toro-Sola MA (1971) Diagnosis of trisomy 22 with quinacrine fluorescence. In: Program and abstracts. Soc Pediatr Res, p 12

Punnett HH, Kistenmacher ML, Toro-Sola MA, Kohn G (1973) Quinacrine fluorescence and Giemsa banding in trisomy 22. Theor Appl Genet 43:134–138

Received August 28, 1980

0340-6717/81/0056/0421/$ 01.00