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Letters to the editors

Di George syndrome and 22q11 rearrangements

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Cytogenetic anomalies of the 22q11 region have sometimes been observed in Di George syndrome (de la Chapelle et al. 1981; Kelley et al. 1982). In such cases some important features of the syndrome may be lacking (due to the precise location of the rearrangement?). We have observed an apparently balanced translocation t(2;22)(q14.1;q11.1) (Fig. 1) in a girl with an incomplete Di George syndrome (telecanthus, microretrognathia, severe aortic coarctation with hypoplastic left aortic arch, decreased E rosettes, and mild neonatal hypocalcemia). The same translocation is present in the normal mother and maternal aunt (Fig. 2). The latter has had her fourth pregnancy aborted because of cardiac and other malformations detected by ultrasound (Di George?). This family may be compared with that published in this journal (Greenberg et al. 1984), in which, among several subjects with a 22pter→22q11.2 deletion some are affected with partial Di George syndrome and others with the complete form.

We would be very interested to hear about similar incomplete Di George syndrome with 22q11 rearrangement for cytogenetic and molecular comparisons.

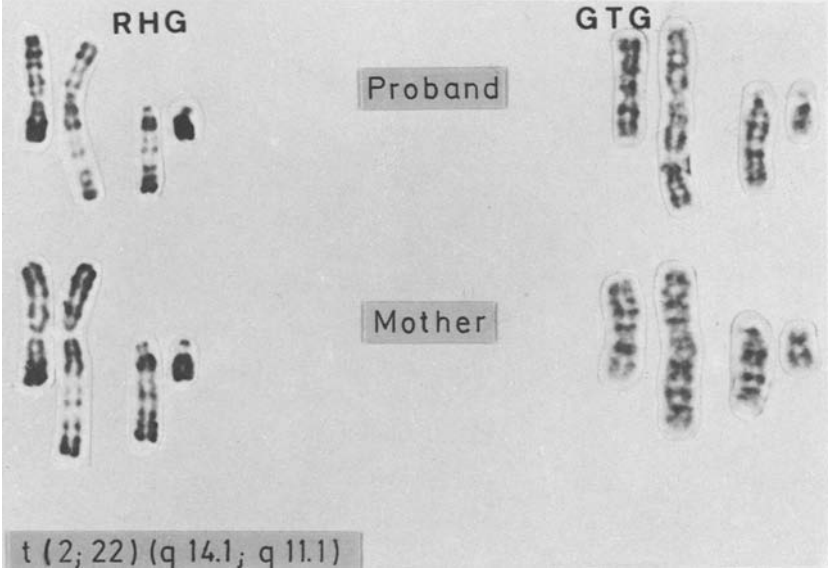

Fig. 1. Chromosomes, RHG and GTG banding techniques. t(2;22)(q14.1;q11.1) without any detectable deletion

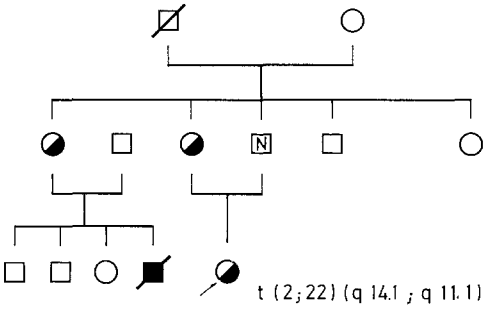

Fig. 2. Pedigree of the family. The proband, her mother, and her maternal aunt have the same balanced translocation

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