couples who have children with Down's syndrome, since their risk of recurrence is considerably greater than that of non-mosaic parent.

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Three generations and seven family members with a t(21q22q) chromosome

Nine patients who had both structural and numerical chromosomal aberrations were found among 275 patients with Down's syndrome. All the structural aberrations were reciprocal translocations. Six presented fresh mutations and three were familial mutations transmitted through several generations. There were five D/G translocations and four G/G translocations.

Before 1972, we did not use banding techniques or autoradiographic methods, so we could not precisely identify the chromosomes in each translocation. In the last two years using the modified banding method of Sumner, Evans, and Buckland (1971) and Wang and Fedoroff (1972), we have been able more precisely to identify three cases of reciprocal translocation. All three belonged to the G/G type; two were 21/22, while one of them was 21/21. One reciprocal translocation 21/22 was familial and the other a fresh mutation.

Our propositus (III.14) and his family represents a remarkably rare familial type of Robertsonian translocation (Hamerton, 1971; Chapman, Gardner, and Veale, 1973). III.14 is the second child of young healthy parents (a 26-year-old mother and a 32-year-old father). The mother (II.15) comes from a large family (Fig. 1). The pregnancy passed normally and delivery was spontaneous at full term. The propositus weighed 3030 g and started to breathe and cry after resuscitation.

Psychomotor development of the propositus was retarded from the very beginning. At 8 months he...
was considerably hypotonic with no interest in his environment. Phenotypically, he had all the important features of Down's syndrome.

Routine cytogenetic analysis of peripheral blood using a modified Moorhead technique, showed a 46,XY,−G,+r(GqGq) karyotype. Using the ASG and trypsin banding techniques the chromosomes in the translocation of the propositus could be precisely identified as No. 21 and No. 22.

The aim was to examine all the members of the propositus' family, beginning with the parents and his sister (III.13). As the father (II.16) had a nor-
mal karyotype, we excluded his family from further analyses. We examined all available members of the family of II.15. II.15 and III.13 were balanced carriers of a t(21q22q), the same as III.14 (Fig. 2).

As shown in Fig. 1, we succeeded in investigating 10 members of the family, representatives of three generations. A grandmother (I.1) her four daughters (II.7, II.11, II.13, II.15), and her five grandchildren (III.9, III.11, III.12, III.13, III.14). The male members of second generation and their descendants could not be examined as they had emigrated. Among the 10 members of the family examined, II.1, II.11, II.13, II.15, III.9, and III.13 were balanced carriers of t(21q22q), while II.7, III.11, and III.12 had normal karyotypes. III.14 was a patient with Down’s syndrome, who besides the structural abnormality of G/G type also had one supernumerary chromosome in group G (Fig. 3).

The small number of families with t(21q22q) reported so far makes this type of reciprocal translocation very interesting from the genetic point of view. The available data seem to suggest an excess of carriers compared to cases with Down’s syndrome but because of the small number of families studied no firm conclusion can be drawn (Hamerton, 1968 and 1971; Chapman et al, 1973).

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REFERENCES

SI Units
With the recommended introduction of SI (Système International) Units for reporting hospital laboratory results, the Editorial Committee of the Journal of Medical Genetics has decided that these will be introduced in the journal from 1975. It has been agreed that where the numerical value of a new unit is the same as at present, the new unit only will be used, eg, haemoglobin: 12 g/100 ml will now be expressed as 12 g/dl. However, where the numerical value expressed in SI Units is different from the value expressed in the units in current use results will, for some time, be reported both in SI and old units, eg, blood pressure: 100 mmHg = 13.3 kPa.

A list of references are given below for the aid of authors and readers and the March 1975 issue of JMG will carry a table of some relevant recommended SI Units.

REFERENCES