that there were male patients with abnormal chromosomal findings. In 1975, we reported on a woman with anosmia and hypogonadism who showed mosaicism 46,XX/47,XX,F?+ (Jones *et al.*, 1975). Like Ventruto and coworkers, we demonstrated an extra chromosome which we 'cautiously' assigned to the F group. Furthermore, our patient pointed to the necessity to examine various tissues as the leukocytic karyotype was 46,XX. Thus, the finding of extra chromosomal material in some of these patients is conspicuous and the question arises whether this is related to the syndrome. Contrary to other women with olfacto-genital dysplasia, our patient was refractory to gonadotropic stimulation (Pergonal). It will be of interest to see what the pertinent testing of Ventruto and coworkers will show.

Yours, etc,

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'Mirror image' chromosome No. 21

Sir,

We wish to report an unusual karyotype found in a recent chromosome survey of 400 subjects with Down's syndrome. The karyotype of a 50-year-old woman with Down's syndrome was found to have a complement of 46 chromosomes with one normal chromosome No. 21 and another longer abnormal chromosome satellited at both ends, best described as a mirror image of chromosome No. 21. The parents of this patient were not available for study and she has a normal sister.

The abnormal chromosome is acrocentric, a little shorter than any of the D group from which it could be distinguished easily in all metaphases examined (Fig. 1a). Giemsa banding showed the abnormal chromosome to be a composite of two No. 21 chromosomes in which the long arms of each chromosome are joined (Fig. 1b).

The appearance of the abnormal chromosome varies with the stage of mitosis. In late metaphase it has the shape of an acrocentric chromosome but in early metaphase the distal ends of the q arms are joined and one or two satellites are clearly seen. Involvement of both ends of the chromosome in satellite association was also seen in many metaphases (Fig. 2).

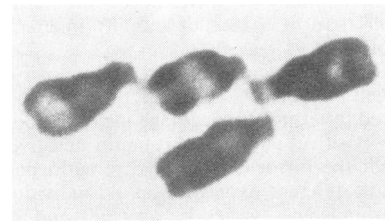

Fig. 2 *Both ends of chromosome in satellite association.*

Similarly abnormal karyotypes in Down's syn-

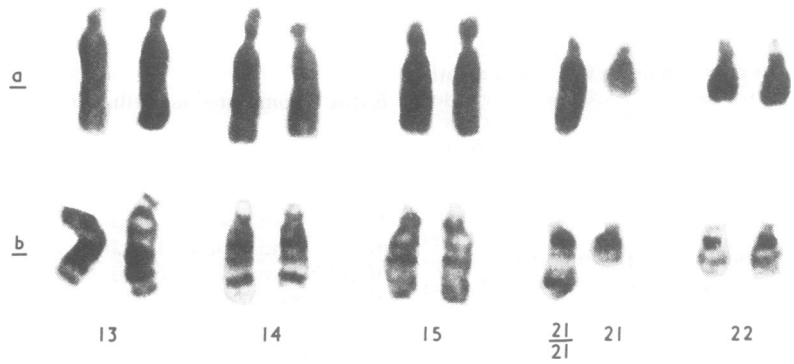

Fig. 1 *D and G groups chromosomes (a) unbanded, (b) Giemsa banded.*

drome have been previously reported as an unusual form of translocation (Zellweger *et al.*, 1963; Warkany and Soukup, 1963; Lejeune *et al.*, 1965). As these cases were reported before the banding technique had been available, the origin of the translocated chromosome could not be determined.

As a result of pedigree studies in two familiar cases of translocation one was shown to be the result of a pericentric inversion of a 21/21 Robertsonian translocation (Soudak *et al.*, 1966) and the other to be a case of reciprocal translocation between chromosomes. No. 21 and 22 with both elements of the reciprocal translocation present in the balanced carriers (Cohen and Davidson, 1967).

Richards *et al.* (1965) reported a subject with Down's syndrome with a similarly unusual chromosome in all cells, but in some metaphases a small acrocentric chromosome was absent. In this case the unusual translocation was considered to be the result of fusion of the long arms of two small acrocentrics following a break and deletion and that it probably arose during parental gametogenesis.

The origin of the unusual chromosome in our case remains debatable. Giemsa banding pattern suggests spontaneous end-to-end adhesion of two No. 21 chromosomes with subsequent loss or inactivation of one centromere. But this requires the unusual occurrence of identical terminal deletions not detectable by Giemsa banding.

It is also possible that sister chromatids have failed to separate at the distal ends of the q arms during the second meiotic division. At anaphase both chromatids, in tandem arrangement, would then migrate to one pole producing a duplicate chromosome No. 21 in one of the two gametes followed by inactivation of one centromere. Fertilization of this gamete could result in a Down's syndrome zygote with 46 chromosomes one of which is a mirror image of chromosome No. 21.

Because the subject has a normal sister and because we have been able to establish with Giemsa banding that the abnormal chromosome is a composite of two chromosomes No. 21, the abnormality must have arisen sporadically.

Yours, etc,

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