has no trace of masculinization nor stigmata of trisomy 18. She is of normal intelligence and has a Turner phenotype, and her dermatoglyphic patterns are also consistent with Turner's syndrome. Thus it appears that the distribution of the cells with the two different karyotypes in the fibroblasts more truly reflects the phenotype than the blood culture findings.

The following reports confirm this assumption. Potter and Taitz (1972) reported monozygotic female twins, one normal female and one phenotypic Turner, both of whom had 45,X/46,XX chromosomal mosaicism in lymphocytes. Fibroblast chromosome analysis revealed pure 46,XX in the normal and 45,X in the Turner twin, and the dermatoglyphics were in accordance with those findings.

A case of 45,X/46,XY/47,XYY with typical Turner's syndrome without gonadoblastoma or trace of masculinization (Roubin *et al*, 1973) had predominantly 45,X cells in cultures from fibroblasts and both gonads, whereas in blood lymphocytes about 50% of cells contained a Y chromosome. This is additional evidence that an XY cell line in about half of lymphocytes does not have a phenotypic influence in X/XY mosaics, if the XY cell line is absent or present only in a low proportion in fibroblast cultures.

Moreover, Beratis *et al* (1972) found a slight mosaicism with 18-trisomic cell line (blood: 5/70, skin: 4/100) in the normal father of a child with trisomy 18. This finding confirms our conclusion that a low proportion of 18-trisomic cells does not necessarily influence the phenotype. Parental trisomy 21-mosaicism of children with Down's syndrome was also reported by Hsu *et al* (1971) and Krmptotic and Hardin (1971).

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Presumptive Mosaic Partial Trisomy Associated with Congenital Anomalies and Mental Deficiency

Summary. The case of a mentally retarded patient with congenital anomalies not typical of any known chromosome unbalance is reported. In his karyotype, 40-6% of the cells were normal, while 59-4% had a missing G and an almost metacentric marker longer than an F chromosome. The abnormal cell line was interpreted as resulting from a chromatid translocation involving the short arm of a No. 22 and a segment from an unidentified chromosome. The translocation probably took place after the first cell division and was followed by segregation of the translocated chromatids. Other obvious hypotheses were excluded by the study of fluorescence patterns. The patient's clinical features may be due to a partial autosomal trisomy.

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A patient with congenital anomalies and mosaicism for an unusual structural chromosome aberration is reported.

Case Report

L.C.V.O., born in February 1965, is the fourth son of healthy, unrelated parents, the mother and father being 43 and 51 years old, respectively at the birth of the propositus. The sibs were clinically normal. There was no history of abortions. In the third month of pregnancy the mother showed psychiatric disturbances, was hospitalized, and treated with electric shock and Amplictil (chlorpromazine, Rhodia), and discharged with slight improvement after two months. The patient was born at term by Caesarean section because of placenta previa; two of his sibs had also been delivered by Caesarean section because of fetal distress. The propositus' birth weight was 3100 g and head circumference was 34.5 cm; he showed deep cyanosis, respiration distress, hypotonia, feeble cry, and unusual facies. Skull radiology, EEG, and fundi were normal. On the third day of life he developed jaundice (total bilirubin = 22.5 mg%; indirect bilirubin = 20.0 mg%) which regressed after two days of phototherapy. The mother was Rh (-), but was direct Coombs (-). Feeding was done through a nasogastric catheter until the third week of life. He was discharged from the hospital at one month of age and from then on had normal weight gain. He held his head up at 10 months, sat at 18 months, and walked at 3 years. Examination at 6 years showed severe mental retardation; he had not developed sphincter control and could not talk. He had moderate generalized hypotonia and was hyperactive. Weight was 21,300 g, height 110 cm (about 10th centile), and occipital-frontal circumference was 55 cm (about 98th centile). He had an impressively large head with high and prominent forehead, and a proportionately large face, flat supraorbital ridges, ocular hypertelorism; mouth breathing, thumb sucking and hypotonia, deformities of face (anteverted nostrils, triangular shaped mouth, mandibular prognathism with malocclusion, elongation of philtrum with a slightly high and somewhat narrowly arched palate); anti-mongoloid slanting of palpebral fissures; the auricles appeared disproportionately small but were normally differentiated; he had a convergent strabismus (left external rectus weakness, also present in two older brothers). There was evidence of perinatal lymphoedema (prominence of hair-follicles on dorsum of hands and fingers);

in addition he showed hyperextensible thumbs ('trigger thumbs'), hypoplasia of both fifth toenails, slight webbing between the third and fourth toes (more pronounced on left than on right), a shallow sacral dimple, and a bilateral pes planus, and calcaneo valgus deformity. The hands appeared short; both showed slight clinodactyly of the fifth digits and a transitional palmar flexion crease.

Dermatoglyphics are summarized in Table I. The Walker index (Walker, 1957) was -4.79. The low total ridge count (TRC) and the ι on the left palm were the only remarkable features.

The rest of the physical examination, fundoscopy, auditory acuity, and results of the EEG were normal.

Cytogenetic Investigations

Chromosome analysis was performed on peripheral blood leucocytes cultured by a modified Moorhead method. The fluorescence banding pattern studies were performed according to the method described by Caspersson, Lomakka, and Zech (1971).

The patient showed a normal karyotype (46,XY) in 39 (40.6%) of the 96 metaphases analysed. In the remaining 57 (59.4%) cells, a G-group chromosome was missing and an almost metacentric marker, larger than chromosomes 19-20 was observed (karyotype: 46,XY, -G, +mar). In most cells the Y chromosome could not be identified (Fig. 1). The patient is, therefore, a mosaic for a normal cell line and a cell line with a structural chromosome aberration. Using fiducial limits of 0.95, the frequency of the abnormal cell line in the blood lies between 50 and 70%.

The fluorescence banding patterns (Figs. 2 and 3) identified the missing G chromosome as a No. 22. The two arms of the marker chromosome differed in pattern; in one of them the pattern was consistent with that of the long arm of chromosome 22, but the pattern of the other arm was not typical enough to allow identification. The other chromosomes as well as those in the normal cells, displayed normal fluorescence patterns.

Both the parents and the sibs of the patient showed normal karyotypes.

TABLE I
DERMATOGLYPHIC PATTERNS

	Digits					TRC	α rd Angle	Palmar Formula	Hallucal Areas
	I	II	III	IV	V				
Right	R	A	U	W	W	105	46°	11.10.8/7.3/2- ι -A ^u .O.O.L.O	L ^d
Left	W	R	U	W	U		70°	9/10.7.6.1- ι r-L ^u .O.O.O.L	W

FIG. 1. F- and G-group chromosomes of (a) the abnormal and (b) the normal cell lines of the proband. The arrow indicates the marker chromosome.

FIG. 2. Fluorescence karyotype of the normal cell line.

FIG. 3. Fluorescence karyotype of the abnormal cell line.

Discussion

The marker chromosome was interpreted as resulting from a reciprocal translocation between a 22 and an unidentified chromosome. This translocation is most likely to have taken place between chromatids in one of the first cleavage divisions. Assuming that the translocated chromatids have segregated, two cells were formed, one with a partial trisomy for the unknown chromosome and monosomy for the short arm of the 22, and the other with monosomy for the unidentified chromosome segment and trisomy for the short arm of the 22. The

first cell gave rise to the abnormal cell line. No cells of the second type were observed; it is assumed that they were unviable or were selected against, at least in the blood.

If this interpretation is correct, a causal relationship between the chromosome aberration and the phenotypic abnormalities may exist. Monosomy for the short arm of the No. 22 probably has no clinical consequence, since carriers of balanced 21/22 translocations of the centric fusion type show a normal phenotype. The partial trisomy present in approximately half of all the cells may have been responsible for the patient's clinical features.

If the translocation had taken place before DNA replication, the chromosome that lost a segment to the No. 22 would be detectable as shorter and/or as showing an abnormal fluorescence pattern.

Trisomy 22 and the possibility of the marker being an isochromosome could be definitely ruled out by the fluorescence banding patterns. Unfortunately this analysis did not disclose which chromosome was involved in the translocation with the No. 22. Since, on the other hand, the clinical features in our patient are not in agreement with any recognized trisomy syndrome, the segment translocated to chromosome 22 remains unidentified.

We think that it is reasonable to conclude that the obvious chromosomal imbalance must have had some deleterious effect on fetal development; however, in view of the patient's gestational and neonatal history, it is not possible to decide which manifestations are due to the chromosome defect and which are due to environmental effects.

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Announcements

The next **Birth Defects Conference** will be held at the Newporter Inn, Newport Beach, California from 16–20 June 1974 under the joint sponsorship of The Harbor General Hospital Campus of the UCLA School of Medicine and The National Foundation March of Dimes. Topics for discussion will include: heritable disorders of connective tissue; epiphyseal dysplasias; mucopolidoses; new malformation syndromes; hypogonadism and genital tract malformation syndromes; new chromosomal syndromes and fetal visualization, and sampling techniques (fetoscopy). Further information can be obtained by writing to: David L. Rimoin, MD, PhD, Harbor General Hospital, 1000 West Carson Street, Torrance, California, 90509, USA.

The **Teratology Society** will hold its 14th annual meeting on 7–10 July 1974 at the Totem Park Conference Center, University of British Columbia, Vancouver, British Columbia, Canada. For further information, write to James R. Miller, PhD, Department of Medical Genetics, University of British Columbia, Vancouver, British Columbia, Canada.