**Case Reports**


Case Report

The propositus is the third daughter of healthy unrelated parents; the father was 38 years and the mother 41 years at her birth. All the family members have normal stature, the older sister being 162 cm, the mother 152 cm, the 21-year-old sister 150 cm, and the 18-year-old sister 160 cm. The patient was born by Caesarean section at 36 weeks; birth weight was 2400 g. Swelling of the dorsum of both feet was noted at birth and persisted for more than one year. The patient was said to have been always smaller than her sisters at comparable ages, but otherwise her physical and mental development was normal.

At 12 years of age she was referred to this hospital because of short stature (length 117-7 cm, weight 25-2 kg, both below third centile) and absence of pubertal signs. She presented the following stigmata of Turner’s syndrome: multiple pigmented naevi, prominent auricles, short broad neck with slight ptérygium, broad chest with widely spaced hypoplastic nipples, relatively large clitoris, hyperextensibility of elbow joints, and short fourth fingers. Radiology of both hands revealed short fourth metacarpals, a coarse trabecular pattern of the bone, and a bone age of 9-10 years, giving a predicted adult height of only 139 cm.

Sex chromatin was studied in buccal smears and in hair root preparations (Schmid, 1967a and b). The chromosomes from blood cultures were studied in

**Turner Phenotype:**

Mosaic 45,X/47,XY,+18*

**Summary.** A 14-year-old girl with Turner phenotype is described, whose lymphocyte and skin fibroblast cultures both revealed a 45,X/47,XY,+18 chromosomal mosaicism. In blood cultures one third and in fibroblasts 7% of the cells had 47 chromosomes. The identity of the Y and the supernumerary 18 were determined by fluorescence and Giemsa banding patterns. The patient is of normal intelligence and does not exhibit any signs of masculinization or stigmata of trisomy 18.

There are several instances of mosaicism with X-monosomic and 21-trisomic cell lines. Cohen and Davidson (1972) published one case of a mosaic 45,X/47,XX,+21, and Prieur et al (1972) one of 45,X/47,XY,+21. Several cases of mosaic 45,X/46,XX/47,XY,+21 (Pfeiffer, 1968) and one case of mosaic 45,X/46,XY/47,XY,+21 (Edgren, de la Chapelle, and Kääräinen, 1966) have been reported. To our knowledge the present case is the first one involving an extra autosomal 18.

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orcein-stained preparations, after staining with quina-
crine mustard (Caspersson, Zech, and Johansson, 1970) 
and after trypsin digestion and Giemsa staining (Sea-
bright, 1971). A fibroblast culture was set up from a 
skin biopsy of the patient. Dermatoglyphics from both 
hands of the proposita were analysed.

Results

X-Chromatin was negative in 500 nuclei 
counted from both buccal smears and hair roots.

Chromosome Studies. The results from 
peripheral blood and a fibroblast culture are shown in 
Table I. Two cell lines were present, one with 
45 chromosomes with a missing C and the other 
with 47 chromosomes including 15 of group C, seven 
of group E, and five of group G, respectively. 
The latter group contained one Y-like chromosome. 
This cell line was found in more than a third of 
lymphocytes but only in 7% of fibroblast mitoses. 
Cells with 46 chromosomes belonged to the 47,XY, +E 
cell line with random losses. The analysis of 
quincarmin-stained preparations showed a brightly 
fluorescent Y in the cells with 47 chromosomes. 
The supernumerary element in group E exhibited 
the characteristics of a No. 18 both in fluorescent 
preparations and in the Giemsa-stained preparations 
after trypsin digestion (Figs. 1 and 2).

TABLE I
CHROMOSOME ANALYSIS

<table>
<thead>
<tr>
<th>Chromosome Count</th>
<th>Total</th>
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<tbody>
<tr>
<td>45* 44* 45 46* 47</td>
<td></td>
</tr>
<tr>
<td>Blood (11.8.1970)</td>
<td>2 2 60 2 34 100</td>
</tr>
<tr>
<td>Blood (3.10.1972)</td>
<td>3 3 92 5 40 100</td>
</tr>
<tr>
<td>Skin fibroblasts (3.10.1972)</td>
<td>3 3 93 7 100</td>
</tr>
</tbody>
</table>

* Random losses.

Dermatoglyphics. There were ulnar loops on 
all 10 fingers. The total ridge count was 142 
(father: 118, mother: 67). Digital triradii a-d were 
present on both sides in normal positions. Palmar 
axial triradii on both sides were in the t position. 
No thenar patterns were present; hypothenar 
patterns revealed ulnar loops on both sides. The 
$atd$ angle was 45° on both sides.

After the finding of chromosomal mosaicism with 
a Y chromosome, laparotomy was performed. A 
hypoplastic uterus with oviducts and gonadal 
streaks in the position of ovaries was found. 
Streaks and oviducts were resected and histo-
logically examined. The streak gonads were found 
to contain neither primordial follicles nor did they 
give evidence of male gonadal elements; the oviducts 
were normal. A psychological examination at that 
time revealed average intelligence but emotional 
infantilism and clinical re-examination showed no 
signs of trisomy 18 in that the face was normal, 
micrognathy absent, and the position of fingers and 
toes not typical of that syndrome. Oestrogen 
therapy was given for a year, and subsequently the 
patient developed an induced regular menstruation 
and scanty pubic and axillary hair. The breasts 
measured 7 and 8 cm, respectively and had darkly 
pigmented nipples and areolae.

Discussion

The most probable explanation for the origin of a 
45,X/47,XY,+18 mosaic is that a 47,XY,+18 
zygote was formed, which lost one Y and one 18 
chromosome during the same early division. The 
age of the mother at birth (41 years) is in favour 
of this hypothesis. Abnormal cell division in a 46,XY 
zygote leading to an 18-trisomic and 18-monomosonic 
cell line (the latter not being viable) and subsequent 
simultaneous loss of a Y and an 18 chromosome is 
more complicated and less likely. It is also rather 
unlikely, but not impossible, that the extra chromo-
some is a structurally rearranged element composed 
of other autosomal or X-chromosomal parts.

All the cases with double aneuploidy, X mono-
somy, and autosomal trisomy mentioned above 
exhibited some features of Down's syndrome. In 
Pfeiffer's case (1968) of a girl with signs of Down's 
syndrome and 45,X/46,XX/47,XX,+21 mosaicism, 
no information about the proportion of the three 
cell lines was given. In the typical Down's syndrome 
reported by Cohen and Davidson (1972), blood 
lymphocytes revealed a 45,X/47,XX,+21 mosaici-
sm in a proportion of 86:101, whereas the skin 
fibroblasts were pure 47,XX,+21. One case of 
45,X/46,XY/47,XY,+21 (Edgren et al, 1966) is 
male and a typical Down's syndrome with very 
pronounced psychomotor retardation; 33 out of 51 
lymphocytes revealed a 21-trisomic cell line and 49 
of 96 were revealed in fibroblasts. On the other 
hand, the case of 45,X/47,XY,+21 (Prieur et al, 
1972), also male, exhibited far fewer signs of Down's 
syndrome and moderate psychomotor retardation. 
Here, from a fibroblast culture, 14 out of 52 cells were 
trisomic, whereas in lymphocytes there were more 
trisomic cells (47 out of 82). In our patient over 
one third of the lymphocytes and 7% of the fibro-
blasts exhibited the karyotype 47,XY,+18; this girl
FIG. 1. Giemsa-stained karyotype containing 47 chromosomes, XY, with a supernumerary No. 18.

FIG. 2. Group E and G chromosomes from two orcein-stained (a and b), one Giemsa-stained (c), and two fluorescent (d and e) mitoses with a 47,XY,+18 karyotype.
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,XXY 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 XY/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 Krmpotic and Hardin (1971).

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REFERENCES

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.