phenotypic effects would suggest that this mis-
division occurred early in development. Trisomic
cells are known to divide more slowly than diploid
cells in vitro (Schneider and Epstein, 1972) and may
explain the lower frequency of trisomic cells in skin
fibroblast cultures in some of the patients reported.

Our second patient had the diagnosis of Kline-
felter's syndrome, presumably as a result of a rou-
tine chromosome analysis. Perhaps a re-evaluation
of moderately retarded patients with this diagnosis
with differential chromosome stains, particularly
if they are known to be mosaics, would identify
additional patients with trisomy 8.

There have been several reports of trisomy 8 in
bone marrow cultures in patients with leukaemia or
related blood diseases (de la Chapelle, Schröder,
and Vuopio, 1972). Changes affecting the C-
group chromosomes are not uncommon in acute
leukaemia, but it is not known yet whether this
involves the No. 8 more often than by chance. The
relationship of the trisomic cell line to the leukaemia
is not known. Follow-up of patients with trisomy 8
will be of interest in this respect.

Addendum

Since submitting this paper, we have seen a third
patient with trisomy 8 mosaicism, a female aged 29
years, who is moderately retarded and has absent
patellae, camptodactyly, and furrows of the soles of
the feet.

The authors would like to thank Dr Helga Muller,
Mrs Cheryl Smith, and Audrey Teng for technical help,
and Mrs Thelma Dietrich, RN, and Mrs Kelly Maher,
RN, for clinical support.

B. F. CRANDALL,* H. N. BASS,† S. M. MARCY,†
M. GLOVSKY,† and C. H. FISH‡

REFERENCES

trisomy mosaicism syndrome. *Heleveca Paediatrica Acta, 27,
281–298.

Caspersson, T., Lindsten, J., Zech, L., Buckton, K. E., and Price,
W. H. (1972). Four patients with trisomy 8 identified by the
fluorescence and Giemsa banding techniques. *Journal of Medical
Genetics, 9, 1–7.

fluorescence d’une trisomie C mosaïque probablement 8: 46,XY/

Prenatal detection of D trisomy

Summary. The second instance of a
prenatally diagnosed fetus of D trisomy is
reported in a 45-year-old woman. The
fetus had bilateral hare lip and cleft
palate, arrhinencephaly, and numerous
other malformations.

Fig. 1. The 22-week fetus with multiple malformations.

Received 8 February 1974.
Recently Butler and his colleagues (1973) reported the first case of 13–15 (D) trisomy to be diagnosed prenatally. We wish to report the second example (Fig. 1).

Case report

A 45-year-old woman was referred for monitoring of her fourth pregnancy, not only because of her age, but also because as well as her two normal children aged 12 and 9, she had a 4-year-old boy with Down's syndrome. In addition, the wife of one of her brothers had a son with Down's syndrome when she was only 23 years old. The woman, her husband, the woman's brother, and sister-in-law were all chromosomally normal, and the two cases of Down's syndrome were of the regular 47 chromosome, trisomic type.

A transabdominal amniocentesis was carried out at about 17 weeks (the last menstrual period was uncertain), after ultrasound placental localization; amnion cell culture and cytogenetics 22 days later showed the fetus to be a female with an extra chromosome in the D (13–15) group. Banding demonstrated it to be an extra 13 chromosome (Fig. 2). A hysterotomy and tubal ligation was performed at about 22 weeks with the delivery within the intact gestation sac of a grossly abnormal female fetus weighing 610 g, and measuring 33.5 cm in crown/rump length, having a bilateral hare-lip, a floating pre-maxilla, a cleft palate, slight micrognathos, an area of scalp aplasia, low set ears, and rocker bottom feet. Internally, there was the mild form of arrhinencephaly (fairly normal cerebral hemispheres and cranial nerves, but with absent olfactory trigones, tracts, and bulbs). A ventricular septal defect, a cystic extra lobe of the left lung consisting of bronchial elements and primitive connective tissue, a Meckel's diverticulum, cystic and dysgenetic kidneys, and a bicornuate uterus were also found.

Discussion

With the increasing number of amniocenteses being carried out at advanced maternal age, principally because of the high risk of Down's syndrome, it is not surprising that chromosome abnormalities other than Down's syndrome are being discovered. So far, apart from these two cases of trisomy D,
A case of partial trisomy 15

Summary. A girl with mental retardation but few distinctive physical abnormalities is described. Chromosome analysis revealed an extra small acrocentric chromosome with both long and short arms satellited which was identified as a deleted chromosome No. 15. The origin of this chromosome is discussed and the clinical findings compared with those of previously reported cases of partial trisomy 15.

Many previous reports of cases with an extra G-like chromosome associated with a non-mongoloid appearance have failed to establish the origin of the extra chromosome. With the development of chromosome banding techniques identification of such chromosomes has been made possible. There have been three recent reports in which an extra small acrocentric chromosome has been identified by banding as a deleted chromosome No. 15 (Magenis et al, 1972; Parker and Alfi, 1972; Bucher et al, 1973).

We present a further case of partial trisomy 15 in which the extra small acrocentric chromosome is satellited at both ends.

Case report

The proposita, born in 1967, is the sixth of seven children of Pakistani parents who are first cousins, their mothers being sisters. The six other children all appeared normal. At the time of the birth the father was 38 and the mother 33 years old. Pregnancy and delivery, at 40 weeks, were normal. Birth weight was 3400 g.

The proposita (Fig. 1) presented at 11 months with delayed development and abnormal limb movements which were convulsive in nature. She had several minor seizures daily. All her developmental milestones had been grossly delayed. She had synophrys and strabismus but no other distinct facial characteristics. The facies were reminiscent, but not diagnostic, of the de Lange syndrome and there was no phocomelia and no

---

Received 5 March 1974.
Prenatal detection of D trisomy

K. M. Laurence, P. J. Gregory and F. Sharp

doi: 10.1136/jmg.11.4.398

Updated information and services can be found at:
http://jmg.bmj.com/content/11/4/398

**Email alerting service**
Receive free email alerts when new articles cite this article. Sign up in the box at the top right corner of the online article.

**Notes**

To request permissions go to:
http://group.bmj.com/group/rights-licensing/permissions

To order reprints go to:
http://journals.bmj.com/cgi/reprintform

To subscribe to BMJ go to:
http://group.bmj.com/subscribe/