Clinical examination revealed a rather small young woman of 4 ft 11 in. in height. Her facies showed typical Crouzon deformities of beaked nose, hypertelorism, moderate ocular proptosis, and maxillary hypoplasia (Fig. 1). The skull showed residual acrocephaly modified by previous cranioplasties. Examination of the back revealed a double primary scoliosis with mild right thoracic and left lumbar rotation. There was a valgus deformity of 10° at the left knee with a left patella subluxing in extension but reducing in flexion. Flexion of the left knee was restricted by 20° compared to the right knee. The right knee appeared normal. Both feet manifested cutaneous syndactyly between the second and third toes. No other clinical abnormality was detected and routine biochemical and haematological investigations were normal. IQ at 15 years was normal.

Radiographs at this stage showed changes typical of Crouzon’s syndrome in the skull and face (Fig. 2). The left knee showed a hypoplastic lateral femoral condyle and the presence of the scoliosis was confirmed (Figs. 3 and 4).

The patient was one of eight children. Examination of the parents and sibs did not reveal any abnormality, and enquiry into the family history did not show any further cases of Crouzon’s syndrome. The parents were unrelated. At the birth of the patient the father’s age was 45, the mother’s 38.

Operation was carried out in September 1971 when the left patellar tendon was transposed medially and distally on the tibia. The patient made an uneventful recovery and when followed up one year after operation had no symptoms of instability in the left knee.

Discussion

This case shows the typical craniofacial manifestations of Crouzon’s syndrome with mild skin syndactyly as described by Saethre (1931) and Chotzen (1932/1933) together with the femoral condyle hypoplasia and genu valgum as described by Temtamy (1966) in Carpenter’s syndrome. The occurrence of scoliosis has so far not been documented as part of any of the syndromes mentioned above.

It is possible that this case represents a fresh mutation with craniofacial characteristics of Crouzon’s syndrome and skeletal abnormalities suggestive of Carpenter’s syndrome together with the mild skin syndactyly described by Saethre (1931) and Chotzen (1932/1933).

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Deletion of the long arms of the Y chromosome with normal male development and intelligence

Summary. Normal male development was found in a man with a proven deletion of the long arms of the Y chromosome. The only phenotypic effect was on his build. This study offers additional proof that all, or most of the long arms of the Y chromosome are not primarily concerned with the determination of male sexual characteristics.

Large deletions of the Y chromosome resulting in small morphologically intact chromosomes with a centromere, give rise to a variety of phenotypic effects which have been described on at least six occasions. Telfer, Baker, and Rollin (1973) reported it in a boy of short stature; Meisner and Inhorn (1972) in a severely retarded man and his normal brother; Nakagome et al (1965) in a severely retarded boy; Muldal and Ockey (1962) described a family with muscular dystrophy and hydrospadias; Vaharu et al (1961) reported a case of gonadal dysplasia in a ‘female’ with an enlarged phallus; and Lo and Kobernick (1965) found a deleted Y cell line in a

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mosaic with a phenotypic intersex. The present case has a deletion of the long arms of the Y chromosome, but he is a phenotypically normal male.

**Case Report**

M.R. was a 23-year-old, unmarried, mature and intelligent, slightly built, timid male of 152 cm, who was otherwise physically unremarkable (Fig. 1). He was said to have always been small as a child. There was no medical history of note except for an unexplained gradual loss of weight of three years duration and at the time of investigation he weighed only 31 kg. There were no organic abnormalities demonstrable except for a spina bifida occulta of S1 and all laboratory investigations including ketosteroid estimations fell within normal limits.

He is the only child of healthy parents, the father was 35 and the mother 33 when he was born. His father is 175 cm and his mother 165 cm tall, two paternal uncles and grandfather are 172 cm, 170 cm, and 170 cm tall, respectively. The patient’s maternal grandfather was 175 cm and a maternal cousin is 185 cm tall.

**Cytogenetic Studies**

Chromosome studies from short-term lymphocyte cultures were carried out on four occasions on the patient and once only on his parents. The cells from the patient contained 46 chromosomes, one of which was a small chromosome the relative size of which was constant and always approximately the same size as the short arms of the G-group chromosomes. On karyotyping, 44 normal autosomes were present and a well-defined X chromosome. It was assumed that the small chromosome represented the remaining portion of a deleted Y chromosome. Banding studies, using a modification of the trypsin G-banding method of Seabright (1971), showed that the autosomes and X chromosome had normal banding patterns (Fig. 2) suggesting that there had been no translocation of the deleted portion of the Y chromosome to another chromosome. Fluorescent studies showed no brightly fluorescing portion of the long arms of the Y chromosome and no Y bodies were discernible in the resting nuclei.

The chromosome constitution of the patient was thus interpreted as 46,XYq−.

Buccal smears from the patient were chromatin negative. Chromosome analyses of the parents yielded normal results. The father had a morphologically normal Y chromosome (Fig. 3).

**Discussion**

The normal male appearance of our patient and the absence of any malformations are undoubtedly due to the normal set of autosomes, the normal X chromosome, and the presence of male-determining genes on the remaining portion of the Y chromosome. The suggestion that these male-determining genes are located on the short arm region of the Y chromosome seems to be supported by the relative normality of our patient and the cases described by McIlree et al (1966) of a dicentric Y chromosome in phenotypically normal males, where a small portion of short arm material was included with the duplicated long arms. Meisner and Inhorn (1972) suggest that the developmentally significant portion of

![Fig. 1. M.R. showing short stature and slight but mature build.](http://jmg.bmj.com/doi/abs/10.1136/jmg.38.4.209)
the Y chromosome is very small, with fertility and height factors on the short arms or close to the centromere on the long arms. Our case and the case of Telfer et al (1973) lend support to this hypothesis, at least in respect to the height factor. In both cases the only phenotypic effect of the deleted Y chromosome is the short stature, which suggests that the height factor is missing. This is reinforced by the fact that the male relatives of our patient are all of average or above average height (Altman and Dittmer, 1972).

These cases, with their short stature, are in

![Fig. 2. Karyotype of a trypsin G-banded cell from the patient.](image)

![Fig. 3. Partial karyotypes of the patient (above) and his father (below) showing the deleted Y and the normal Y chromosome, respectively.](image)
Case reports

marked contrast to the large stature commonly
seen in XYY males, where, there is a duplication of
each active factor carried on the Y chromosome.

We wish to thank Dr Byron Evans, Consultant
Physician, University Hospital of Wales (Cardiff)
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A 48,XXXX female

Summary. A four-year-old mildly
retarded girl with a 48,XXXX karyotype
is described. Her phenotype is com-
pared to previously reported cases.

The first tetra-X female was described by Carr,
Barr, and Plunkett (1961). Since then only 16
other cases have been reported. The present paper
reports a further case and the phenotype and origin
of this abnormality are discussed.

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Case Report

The proposita was a Caucasian female born in August
1969 as the 2550 g product of an uneventful pregnancy
and term delivery to a 25-year-old mother. Her
mother, father, and the two older sisters were healthy
and had normal intelligence and stature. During the
first 6 months of life she had feeding difficulties, with
frequent regurgitation and vomiting. Her growth and
development were slow and at 32 months she was ad-
mitted to the Children's Centre in Winnipeg for inves-
tigation. Physical examination at that time showed a
height of 85 cm, weight of 9.2 kg, and a head circum-
ference of 43.5 cm, all of which were below the third
percentile for her age. Facial features included pro-
minent eyebrows growing together in the midline
(synophrys), small epicanthic folds, and a small mandible
(Fig. 1). She had a high arched palate. Her hands
showed bilateral simian creases. Her gait was unsteady
and she was mildly hypotonic. The remainder of her
physical examination fell within normal limits. Labora-
tory tests showed her to have normal levels of calcium,
phosphorus, alkaline phosphatase, BUN, glucose, and
thyroxine. Radiology showed a bone age of 12 months
and a reduced craniofacial ratio. Re-examination at 44
months did not reveal any other abnormalities and her
measurements were still below the third percentile.

Developmental Assessment (E.L.). Evaluation
at 45 months showed her to be functioning for the most
Deletion of the long arms of the Y chromosome with normal male development and intelligence
Helen Langmaid and K. M. Laurence

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