an interstitial duplication of the 7q22→q32 region in
cases with frontal bossing, strabismus, large wide
ears, and hypotonia. Turleau et al proposed two
syndromes based on these distinctive characteristics.
More recently, Schmid et al suggested a third
syndrome as their patients had asymmetry of the
skull, small dysplastic ears, kyphoscoliosis, and
seizures, as well as strabismus and hypotonia.

Despite apparent specificity there is some pheno-
typic overlap and certain common features are
described in all cases. These include mental retard-
ation, low set ears, hypertelorism, small palpebral
fissures, a short upper lip, and a short neck.

Almost all reports of partial trisomy 7q result from
a familial reciprocal translocation or insertion trans-
location. In addition to trisomy, partial monosomy
of a variety of other chromosomes is represented.

There are also reports of derivative chromo-
somes resulting from pericentric inversions of
chromosome 7. The first de novo report was by
Wahrman et al. A very large chromosomal segment
was duplicated and involved the 7q11→qter region.
This was also the first case reported with genitouri-
nary and renal anomalies. Our patient is similar
clinically in that he had an atrophic kidney, a
ureteral anomaly, and cryptorchidism, as well as the
similar facial features that are characteristic of 7q
trisomy. The specificity of renal anomalies is further
illustrated by Ramirez and Uribe as they report full
trisomy 7 in an infant who presented with Potter’s
syndrome. The pathological examination revealed
an absent left kidney and a right polycystic kidney.

Although a gene dosage effect could not be
demonstrated by enzymatic assay or blood group
linkage analysis, high resolution banding suggested
a duplication of the 7q11→q22 region as compared to
the other 7q bands. The phenotypic similarities to
full and partial trisomy 7 make it unlikely that the
extra material is derived from another chromosome.
The most likely mechanism producing the derivative
chromosome is three breaks resulting in an insertion
of the region in question or an unequal cross over in
meiosis.

We wish to thank W L Marsh at the New York
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B-glucuronidase enzyme assay.

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De novo interstitial direct duplication of 15q: 46,XY,dup(15)
(pter→q24::q14→q21·1::q24→qter)

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SUMMARY A profoundly retarded, slightly dys-
morphic male was re-examined cytotegenetially
by high resolution GTG banding and found to
have a de novo interstitial direct duplication of
15q.

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Case report

The patient reported here (fig 1) is a 12-year-old
Caucasian male, the product of a 41-week gestation
of a 34-year-old gravida 2 para 2 woman and her
36-year-old non-consanguineous husband. During
the eighth and ninth months of pregnancy, hydro-
chlorothiazide (Hydrodiuril) for significant
dependent oedema was used three times a week, with dietary salt restriction. The pregnancy was otherwise without complication and a spontaneous vertex delivery was achieved following a labour of approximately 3 hours. Oxygen and mucous aspiration were required to attain an Apgar score of 8 at 5 minutes. Birth weight was 2670 g and the child was noted to have cryptorchidism.

At 4 months of age chromosome analyses were done on peripheral blood from the proband and his parents, but no abnormality was detected. At 8 months a right orchiectomy was performed because of impaired cord blood supply. At 18 months the patient had a left orchiopexy and received a prosthetic implant (ultimately extruded spontaneously). Shortly after his surgery, he was referred for orthopaedic consultation because of skeletal problems with his back and feet; these were treated symptomatically. An infant-parent stimulation programme was suggested for his developmental retardation following assessment. Psychological evaluation at 22 months indicated a mental age of 10 months, with an IQ of 47 on the CIIS.

We continued to follow the patient at 6-month to yearly intervals. When he was 7 years old chromosome analysis (which had previously been done elsewhere) was repeated. Again, no structural abnormalities were detected in Giemsa banded metaphase cells. Following a seizure at the age of 11 years, EEG, thyroid and growth hormone assays, and other laboratory studies were done. With the exception of the EEG, which was compatible with a seizure disorder, the tests were normal and the diagnosis remained 'syndrome of unknown aetiology'. At 12 years, by which time a clinical geneticist had been added to the staff, it was felt that syndrome identification should be re-explored, and a genetics consultation was requested.

Physical examination revealed short stature, although the exact height could not be measured due to hyperactivity. Head circumference of 49.1 cm was below 2 SD for age. The face was distinctive, with a narrow forehead, high nasal bridge, narrow palate, dental malocclusion, small ears, and micrognathia. Additional findings included lumbar kyphosis, bell-shaped chest, mild scoliosis, scrotal hypoplasia, and micropenis (2.5 cm long, 1.5 cm wide). There was no cardiomegaly and no thrills or murmurs were heard. The abdomen was protuberant but could not be adequately examined for organomegaly owing to poor cooperation. Mental retardation was profound and intelligible speech was absent.
Case reports

A repeat cytogenetic analysis, using high resolution GTG banded prometaphase chromosomes,1 revealed an interstitial direct duplication in the long arm of chromosome 15 (fig 2). We interpret the karyotype as 46,XY,dir dup(15) (pter→q24::q14→q21·1::q24→qter), in accordance with the International System for Human Cytogenetic Nomenclature.2 The proband is therefore trisomic for a small interstitial segment of 15q. Repeat studies of the parents confirmed 46,XX and 46,XY karyotypes.

Discussion

A number of reports have been published on partial trisomy of 15q resulting from parental translocation or inversion. Yip et al3 compared eight such cases with a ninth case of de novo tandem duplication. In all but one,4 the most proximal breakpoint was q21 and all were trisomic to qter. The case reported here is only the second de novo case observed and the first 15q interstitial direct duplication. It is of interest that although the trisomic segment in our case is more proximal than in the majority of the other cases, the physical findings are very much in agreement.

While we would not advocate high resolution banding as a routine procedure nor suggest that every suspect patient with a normal karyotype be re-analysed, it would seem prudent to apply this newer technique in certain special instances. More cases with minor structural abnormalities are obviously needed before a precise picture of 15q trisomy evolves.

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De novo interstitial direct duplication of 15q: 46,XY,dir dup(15) (pter leads to q24::q14 leads to q21 X 1::q24 leads to qter).

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