A case of 47,XY,+der(15),t(3;15) (p25;qll)pat presenting as partial 3p trisomy syndrome with multiple joint contractures

SUMMARY Partial 3p trisomy is a rare chromosomal syndrome which results in a characteristic phenotype. We present a case of partial 3;15 trisomy with clinical features of partial 3p trisomy syndrome and multiple joint contractures.

Partial 3p trisomy syndrome was described originally by Rethoré et al. Since her original description of three cases, seven additional cases, characterised by varying degrees of trisomic material for the short arm of chromosome 3, have been reported. Aarskog described a child with duplication 3, deletion 18q with the clinical features of partial 3p trisomy syndrome. A female sib born after that publication was found to be similar phenotypically, and on chromosome analysis had duplication 3p, deletion 18q. These children probably represent two additional cases of this rare chromosomal syndrome. We describe a patient with the clinical stigmata of partial 3p trisomy syndrome and multiple joint contractures, who was found to have an unbalanced translocation thought to represent a partial trisomy for the short arm of chromosomes 3 and 15 and a small segment of the proximal long arm of chromosome 15.

Received for publication 5 December 1979

---

Case report

The proband was a term male born to a 39-year-old gravida 3, para 1, aborta 1, blood type A positive mother. Decreased fetal movements were noted in the last trimester and there was rapid weight gain. Based on the ultrasonographic findings of polyhydramnios in the third trimester, a caesarean section was performed when spontaneous labour occurred. One and five minute Apgar scores were 5 and 7, respectively.

At birth the baby's length was 49.5 cm (10th to 25th centile), weight was 2900 g (10th to 25th centile), and head circumference was 37.75 cm (95th centile). There was frontal bossing with temporal narrowing. The facies was square shaped with prominent cheeks. There were downward slanting palpebral fissures and hypertelorism. The ears were low set. The philtrum was elongated. The mouth was large with down-turned corners. The palate was normal and there was a mild degree of retrognathia (fig 1). No murmur or organomegaly were detected. The genitalia were hypoplastic with laterally undescended testes. There were joint contractures of the elbows, wrists, hips, and knees, and camptodactyly of the fingers and toes. The infant was lethargic with generalised hypotonia. The suck root, and Moro reflexes were absent.

Nasogastric feeding was started as a result of sucking difficulties. On the third day of life seizure

---

**FIG 1** Patient with partial 3p trisomy syndrome at 2 days of age. Note elbow and finger contractures.
activity was observed and phenobarbital was started. A CT scan disclosed slight enlargement of the ventricular system with no other structural abnormalities. A chromosome analysis was obtained (see Chromosome findings). After 4 seizure-free weeks, phenobarbital was discontinued. The patient became more alert and began to tolerate oral feeds. He was discharged at 7 weeks of age. At 4 months of age his growth parameters were unchanged. He continued to have generalised hypotonia with significant developmental delays in all areas as assessed by the Denver Developmental Screening Test.

**Chromosome findings**

Chromosome analysis was performed on peripheral blood from the proband using a modification of the method of Moorhead et al. The cells were G banded and then examined microscopically. Of 20 cells, 100% contained 47 chromosomes with a small extra marker chromosome. In order to determine the origin of this marker, a karyotype was performed on the proband's parents. The proband's father had a balanced reciprocal translocation between chromosomes 3 and 15. His karyotype was 46,XY,t(3;15) (p25;q11) (fig 2a). The mother's karyotype was 46,XX. It was concluded that the proband's karyotype was 47,XY,+der(15),t(3;15) (p25;q11)pat (fig 2b). The marker chromosome represented a 3;15 unbalanced translocation of paternal origin. The proband was trisomic for the distal short arm of chromosomes 3 and 15 and a small segment of the proximal long arm of chromosome 15. The sister also had the balanced reciprocal 3;15 translocation of paternal origin.

**Discussion**

This patient is the first reported case of partial 3;15 trisomy to present with many of the characteristic phenotypic features of partial 3p trisomy syndrome (table). The chromosomal abnormality was the result of a balanced reciprocal translocation of paternal origin. Three cases in addition to our patient presented with hypotonia. Low set ears and downward slanting palpebral fissures, which were found in our patient, have not been described previously. Although five other cases also had partial deletions of the reciprocal chromosome, no significant phenotypic differences were noted. There have not been any reported cases of partial 3p trisomy syndrome with partial trisomic material for the reciprocal chromosome. Additional chromosome 15 material was identified on our patient's karyotype. Additional 15 material has not been consistently associated with specific dysmorphology. Although mental retardation appears to be the common denominator in cases of partial trisomy 15 syndrome, low set ears, micrognathia, downward slanting palpebral fissures, had been described in a number of cases. We, therefore, cannot ignore the potential influence of the additional 15 material on our patient's phenotype.

This patient was found to have multiple joint contractures and camptodactyly of the fingers and toes at birth, a feature not previously described in partial 3p trisomy syndrome. However, three other cases of partial 3p trisomy syndrome were found to have camptodactyly of either the fingers or toes and of these, one had contractures of the wrists and elbows as well. While our patient's joint findings may represent an additional less frequently associated clinical feature, this observation may be an

**Table Clinical findings in partial 3p trisomy syndrome**

<table>
<thead>
<tr>
<th>Feature</th>
<th>Previous cases</th>
<th>Present case</th>
</tr>
</thead>
<tbody>
<tr>
<td>Microcephaly</td>
<td>4/10</td>
<td></td>
</tr>
<tr>
<td>Frontal bossing</td>
<td>8/8</td>
<td>+</td>
</tr>
<tr>
<td>Temporal narrowing</td>
<td>5/8</td>
<td>+</td>
</tr>
<tr>
<td>Square facies</td>
<td>9/10</td>
<td></td>
</tr>
<tr>
<td>Prominent cheeks</td>
<td>8/8</td>
<td></td>
</tr>
<tr>
<td>Hypertelorism</td>
<td>10/11</td>
<td></td>
</tr>
<tr>
<td>Epicanthus</td>
<td>8/10</td>
<td></td>
</tr>
<tr>
<td>Down-turned corners of mouth</td>
<td>9/10</td>
<td>+</td>
</tr>
<tr>
<td>Prominent philtrum</td>
<td>9/10</td>
<td>+</td>
</tr>
<tr>
<td>Retrorgnathia/micrognathia</td>
<td>8/11</td>
<td></td>
</tr>
<tr>
<td>Short neck</td>
<td>7/8</td>
<td></td>
</tr>
<tr>
<td>Congenital heart disease</td>
<td>10/12</td>
<td></td>
</tr>
<tr>
<td>Gastrointestinal malformation</td>
<td>4/8</td>
<td>-</td>
</tr>
<tr>
<td>Renal malformation</td>
<td>2/4</td>
<td></td>
</tr>
<tr>
<td>Hypoplastic male genitalia</td>
<td>6/9</td>
<td>+</td>
</tr>
<tr>
<td>Mental retardation</td>
<td>4/4</td>
<td>+</td>
</tr>
</tbody>
</table>

**FIG 2** Partial karyotypes of two cells of the father (a) and the proband (b). Breakpoints in the father's cells are indicated by arrows.
incidental finding. It is anticipated that accumulation of data from newly reported cases will help determine whether a possible association between joint contractures and partial 3p trisomy syndrome exists, or whether these clinical manifestations may, in part, be the result of the presence of additional 15 material.

We wish to thank Drs Allan Bloom, Nuhad Dinno, Bernard Weisskopf, and Frank Yen for their helpful comments.

JOSEPH H HERSH, ROBERT M GREENSTEIN, JANICE C PERKINS, AND PATRICIA C REARDON
University of Louisville, School of Medicine, Child Evaluation Center, Louisville, Kentucky; and Department of Pediatrics, University of Connecticut Health Center, Farmington, Connecticut, USA

References


Requests for reprints to Dr Joseph H Hersh,
University of Louisville, School of Medicine, Child Evaluation Center, 334 E Broadway, Louisville, Kentucky 40202, USA.

De novo duplication 1q32-q42: variability of phenotypic features in partial 1q trisomies

SUMMARY A de novo tandem duplication of 1q32—q42 was observed in a 7-month-old mentally retarded and malformed male infant.

Karyotype-phenotype correlation in other similar unbalanced trisomies has shown psychomotor retardation, micro- or retrognathia of both, and low set or malpositioned ears to be the most common features associated with this newly recognised syndrome. However, after reviewing patients with duplication of regions 1q2, 3, and 4 and 1q2 and 3, it was concluded that similar non-specific clinical features are also present in these 1q imbalances.

On the whole, a rather wide range in phenotypic expression has been observed in different cases. Thus it is concluded that, at present, it is impossible to delineate the profile of the syndromes resulting from partial 1q trisomies.

A number of structural variations and anomalies of chromosome 1 have been reported.1 Among duplications of the long arm (1q+), the 1qh variant is the most common.2 3 The enlargement of chromosome 1q is in these instances the result of an elongation of the heterochromatic secondary constriction (h).

Acquired anomalies of chromosome 1 are found in 16·2% of malignant disorders, most of which are full or partial trisomies.4 5

Among congenital anomalies of the long arm of chromosome 1, 25 partial 1q trisomies have been reported, most of which were segregating from parents heterozygous for balanced reciprocal translocations involving the long arm of chromosome 1q (Vianello, 1979, personal communication).4 18 In six patients, partial duplication 1q occurred as a de novo mutation.18 24 The clinical picture in these patients is variable.

We report a 7-month-old infant who came to our attention. Karyotype-phenotype correlations are attempted on data derived from 16 cases of partial 1q imbalances.

Case report

The proband was the product of the second pregnancy of a 20-year-old mother and a 25-year-old father. The parents were healthy and unrelated. A