Monosomy 10qter: a new case

Michelle Teyssier, Christiane Charrin, Jacqueline Dutruge, Christophe Rousselle

Abstract

A new case of terminal deletion 10q26-qter is described. The phenotypic features are compatible with those of the previously reported cases. Deafness is reported for the first time.

Figure 1 The patient at 4 months.

Laboratoire Central
D'Hématologie et de
Cytogénétique,
Hôpital Edouard
Herriot, Place
d'Arsonval, 69374
Lyon Cedex 03,
France.
M Teyssier
C Charrin

Laboratoire
d'Histologie-Embryo-
logie-Biologie de la
Reproduction, Faculté
de Médecine
Lyon-Nord, 8 Avenue
Rockefeller, 69373
Lyon Cedex 08,
France.
M Teyssier

Clinique Médicale
Infantile, Centre
Hospitalier Lyon-Sud,
69310 Pierre-Benite,
France.
J Dutruge
C Rousselle

Correspondence to
Dr Teyssier, Laboratoire
d'Histologie-Embryologie-
Biologie de la Reproduction,
Faculté de Médecine
Lyon-Nord, 8 Avenue
Rockefeller, 69373 Lyon
Cedex 08, France.

Received 13 August 1991.
Accepted 27 August 1991.

Case report

The proband, a male infant, was born by normal spontaneous vaginal delivery at 40 weeks' gestation after an uncomplicated pregnancy. He was the second child of healthy, unrelated parents, a 32 year old father and a 33 year old mother. Birth weight was 3030 g and Apgar scores were 3 at one minute and 7 at five minutes. Intubation was initially required but subsequently respiration was satisfactory.

Clinical examination showed severe axial and moderate segmental hypotonia, scaphocephaly, many dysmorphic facial features (deep set eyes, downward slanting, short palpebral fissures, hypertelorism, left epicanthus, broad nasal bridge, malformed, low set ears, and retrognathia), a short neck, widely spaced nipples, cubitus valgus, small penis and scrotum with undescended testes, and normal extremities (no syndactyly or clinodactyly). The cry was abnormal.

Laboratory investigations (echocardiogram, electrocardiogram, cranial and abdominal ultrasound, cerebral scan, and digestive system) were normal and the kidneys, bladder, liver, spleen, gall bladder, and pancreas showed no anomalies.

Clinical and neurodevelopmental evaluation was undertaken at regular intervals. Weight and height progressed normally but psychomotor retardation was soon evident. At 4 months

of age, there was no microcephaly but the face appeared asymmetrical with frontal bossing more prominent on the left, right midface hypoplasia, and all the dysmorphic features already described (fig 1). There was severe axial hypotonia (the infant could not raise his head) and segmental hypertonia. Auditory tests showed significant bilateral deafness.

Chromosome analysis was performed on peripheral lymphocytes. R banding showed a terminal deletion of chromosome 10 with the breakpoint in q2-6 (fig 2). Chromosome analysis of both parents was normal. A lymphoblastoid cell line is available from this patient, which is banked at CIRC, 150 Cours Albert Thomas, 69008 Lyon, France (ID No IARC 1719).

Discussion

Deletions of the long arm of chromosome 10 are rare. Nine patients with monosomy for the chromosome region 10q26-qter have been described previously: seven with a de novo deletion,^{1,2} one with a duplication of 8q24.3 (chromosome derived from a maternal translocation),³ and one with a small deletion of chromosome 15 (45,XY,-10,-15,+der(10)t(10;15)(q26;q13).⁴ In addition, six reports describe deletions with breakpoints at 10q25 or 10q23.^{1,5}

An unusually large number of the patients had anoxia or respiratory distress at birth, low birth weight, hypotonia, and some dysmorphic craniofacial features. The children displayed severe growth and mental retardation and strabismus. Many features observed in our patient are similar to those reported in previous cases including deep set eyes, short palpebral fissures, hypertelorism, malformed and low set

Figure 2 The patient's chromosomes 10 from three metaphases (R banding). The deleted chromosome is on the right.

ears, broad nasal bridge, small nose, short neck, cryptorchidism, mental retardation, and an abnormal cry. However, in contrast to previous publications, we did not note low birth weight, growth retardation, or microcephaly. Congenital heart defects, anomalies of the kidneys and urinary tract, and anomalies of the hands and feet (syndactyly), which have occasionally been reported, were not found. Our proband had midface hypoplasia, like the case of Smith *et al.*⁴ Hypotonia has frequently been reported, but Gorinati *et al.*¹ described hypertonia and hyperreflexia; in our case, there was severe hypotonia of the head and back and scapular and ischiotibial hypertonia so the child's movements were cramped. The association of deafness and monosomy 10qter, observed here, has not been described before.

Several features are common to the majority of reported cases. Others, such as urinary tract anomalies,¹ cystic kidney,⁴ and deafness (our case) are less frequently described. However, with so few cases of 10qter chromosomal abnormality reported, it is too early to know if a definitive clinical syndrome is associated with the chromosome deletion.

- 1 Gorinati M, Zamboni G, Padoin N, Dodero A, Caufin D, Memo L. Terminal deletion of the long arm of chromosome 10: case report and review of the literature. *Am J Med Genet* 1989;33:502-4.
- 2 Mehta L, Duckett DP, Young ID. Behaviour disorder in monosomy 10qter. *J Med Genet* 1987;24:185-6.
- 3 Shapiro SD, Hansen KL, Pasztor LM, *et al.* Deletion of the long arm of chromosome 10. *Am J Med Genet* 1985;20:181-96.
- 4 Smith A, Evans WA, Woolnough H. Post-mortem studies on two patients with 1-2 band cytogenetic deletions: 10q26-qter and r(9)(p24q34). *Ann Genet (Paris)* 1989;32:220-4.
- 5 Vanlieferinghen P, Dechelotte P, Charbonné F. Délétion partielle 10qter. Une nouvelle observation. *Ann Genet (Paris)* 1987;30:118-21.