Ecaruca Patient of the Month

Chromosome aberration
47,XY,+der(21)t(1;21)(q31;q10)mat
Partial trisomy 1q, region 1q31→qter

Patient
A one year and 9 months old boy with mild dysmorphic features, moderate psychomotor retardation, pre-axial polydactyly and a ventricular septal defect.

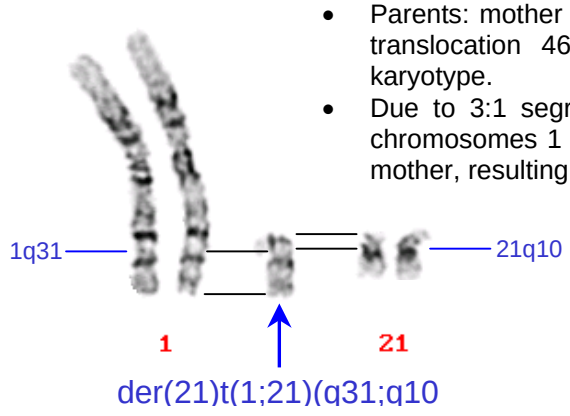
Background information
Partial trisomy of 1q is a rare condition. The reported cases include trisomies of all segments of the long arm of chromosome 1, but most consist of trisomy 1q32-qter. Mostly, the 1q trisomies are accompanied by other chromosomal abnormalities which make it difficult to define a phenotype. Six cases of pure distal 1q duplications have been reported.
Although the short arm of chromosome 21 is also duplicated, this has no clinical significance because this region carries almost no euchromatic material. Therefore, this patient has an almost 'pure' partial trisomy 1q.

Clinical details

- Last age of examination: 1 year and 1 month
- Obstetric history mother (G3P2A1): one healthy daughter, one intra-uterine fetal loss at 20 weeks of gestation
- Perinatal history : He was born after 42 weeks of pregnancy, with a birth weight of 3,960 g (+ 0.5 SD) and he developed respiratory distress for which he received treatment.
- Congenital anomalies: He was born with a pre-axial polydactyly of the left thumb (2 distal phalanges), long nails of fingers and toes and a small ventricular septal defect.
  Ultrasound examination of the cranium, abdomen and kidneys did not reveal any abnormalities.
- Psychomotor development: at age 1 year he could reach, and at 1 years and 8 month he could sit independently. Speech development is delayed. Hearing and vision are normal.
- Growth parameters at 13 months: he had a length of 72.5 cm (-1.5 SD), a weight of 8,480 g (-1.5 SD) and a head circumference of 47.5 cm (0 SD). His facial features included a prominent metopic ridge, an open anterior fontanel, epicanthic folds, a long philtrum and angular ears. Furthermore, he presented with axial hypotonia.
- Treatment: at the age of 10 months he had a circumcision because of phimosis. Furthermore, he frequently receives treatment (antibiotics) for recurrent respiratory infections.
- He attends physiotherapy and speech training and visits the cardiologist regularly. At the age of 3 years he will be operated to correct the polydactyly of the left thumb.

Cytogenetic details:

- Routine GTG banding: 47,XY,+mar in all examined cells.
- FISH: analysis with centromere probes specific for the acrocentric chromosomes, revealed that the marker chromosome was positive for the centromere probe specific for chromosomes 13 and 21.
- Parents: mother appears to be a carrier of a balanced chromosomal translocation 46,XX,t(1;21)(q31;q10). Father has a normal male karyotype.
- Due to 3:1 segregation at meiosis, the patient inherited a normal chromosomes 1 and 21 plus the derivative chromosome 21 from the mother, resulting in a partial trisomy 1q31qter.



Summary of some other patient with trisomy 1q

	Machlitt *et al* (2005)	Nuño-Arana *et al* (2001)	Malgorzata *et al* (2003)	Kimya *et al* (2002)	Present patient
Duplicated region	q21-qter	q32-qter	q32-q44	q32-q44	q31-qter
Sex	male	male	male	female	male
Age	23 weeks of gestation termination of pregnancy	newborn (died age 12 days)	7 months (died age 9 months)	22 weeks of gestation termination of pregnancy	20 months
Mental retardation	unknown	unknown	+	unknown	+
Pre-axial polydactyly	+	-	-	+	+
Macrocephaly	+	-	-	+	-
Facial features					frontal bossing epicantic folds
Kidney abnormality	+	+	+	+	-
Heart defect	+	+	+	-	+
Other	bilateral microphthalmia left persistent vena cava sup omphalocele urethral obstruction duodenal obstruction	absent gall bladder	11 pair of ribs		phimosis axial hypotonia

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