European Cytogeneticists Association
Register of Unbalanced Chromosome Aberrations

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ECARUCA Presentations

Case of the Month

Chromosome aberration

Deletion 13q14.3 to q21.32

Patient

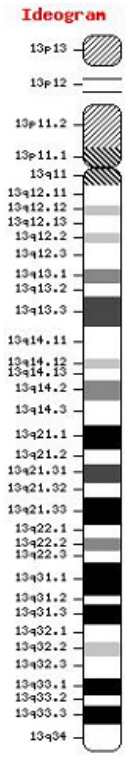
A 4-year-old boy with mild clinical features; a large head circumference, moderate motor developmental delay and introvert behaviour.

Background information

Distal 13q deletions have been described relatively often. However, interstitial deletions in the region 13q14-q21 are rare; three patients have been published. All three have mild clinical features and are (mildly) mentally retarded.

Clinical details

- Last age of examination: 4 years and 3 months.
- Perinatal history: unremarkable, birthweight 3620 grams at 38+3 weeks (50th centile).
- Congenital anomalies: mild micrognathia, dysmorphic ears and camptodactyly of the second finger of the left hand. Ultrasound examination of cranium, abdomen, scrotum and hips did not show any abnormalities.
- Psychomotor development: sitting and walking without support at 7 and 18 months respectively. Due to hypotonia and increased joint laxity his motor skills were inadequate for age. Speech development was normal, however information handling was slow. Hearing and vision were normal.
- Dysmorphic features at 4 years and 3 months: mild overgrowth with a height of 113 cm (+2,0 SDs), head circumference of 55 cm (+2,0 SDs) and mild obesity with a weight of 25.2 kg (+2,0 SDs corrected for height). The facial features included dolichocephaly, epicanthic folds, slightly over folded ear helix and a short philtrum.
- Behaviour: Introvert; he had good contact with his parents, but was reluctant to play with other children of his age.
- Treatment: The camptodactyly was conservatively treated with a splint with good results. Physiotherapy showed an improvement of the motor development, although the level of performance was still delayed.



Cytogenetic details

- Routine GTG banding: 46,XY,del(13)(q14.3q21.32) in 25 metaphases.
- FISH: Whole Chromosome Paint of chromosome 13 showed fluorescent signals on the normal and deleted chromosome 13 only, indicating that no other chromosome was involved. Additional FISH studies with the RB-1 probe showed a signal on both chromosomes 13, thus excluding a deletion of the retinoblastoma region.

Parents: Both parents had a normal karyotype.

Summary of all known patients with a deletion 13q14-q21

Feature	Present case	Noel *et al*	Brown *et al*	Brown *et al*
Deleted region	q14.3-q21.32	q14-q22	q14.3-q22	q14.3-q22
Sex	Male	Male	Female	Female
Age (years)	4	15	54	4.5
Retardation	Mild	Moderate	+	+
Stature	Tall	Short	Unknown	Short
Congenital malformations	Camptodactyly	Microphthalmia	Renal agenesis	Renal agenesis Heterochromia of iris

Striking features

Although the deletion comprises a region of ~ 16 Mb, the clinical features are remarkably mild.