PARTIAL DUPLICATION (1)(P22.1P31.1) 
REPORT ON A BOY WITH MENTAL RETARDATION, 
ABNORMAL GENITALIA AND ABSENT PATELLAE

Valerica Belengeanu, Kinga Rozsnyai, Cristina Gug, Alina Belengeanu, Simona Farcas

REZUMAT
Este descris cazul unui băiat de 7 ani cu o duplicație parțială a bratului scurt al cromosomului 1, o situație raportată foarte rar în literatură, care pe lângă manifestările comune asociate cu această anormalie cromozomială, ca retard de creștere intrauterin, microcefalie, retard mental și criptorhidie, prezintă și absența bilaterală a patelelor.

Cuvinte cheie: duplicație parțială 1p, trisomie parțială 1p, absența patelelor

ABSTRACT
We describe a 7 year-old boy with a partial duplication of 1p, a finding very rarely reported in the literature. Besides the common features associated with this chromosomal abnormality, such as: intrauterine growth retardation, microcephaly, mental retardation and cryptorchidism, the patient also presented bilateral absence of patella.

Key Words: partial duplication 1p, partial trisomy 1p, absent patella

INTRODUCTION
There are few reports of partial duplications of the short arm of chromosome 1, almost each of them with different chromosomal breakpoints, thus presenting with varying clinical manifestations, that include low birth weight, microcephaly, cleft/high-arched palate, mental retardation as common features, and variable additional findings, as minor facial anomalies, brachydactyly and fifth finger clinodactyly.

CLINICAL REPORT
A. H. is the only child of unrelated, healthy parents, born after a full term, uneventful pregnancy and normal delivery, with a birthweight of 2500 g. The early childhood period was complicated with repeated respiratory tract infections.

On clinical examination at the age of 7 years, he had normal height (123 cm, ≈ 50th centile) and weight (24 kg, ≈ 50th centile). His head circumference was 49.6 cm (< 3rd centile). He had short, horizontal palpebral fissures, bilateral ptosis more pronounced on the left, left eye esotropia, wide and prominent nasal bridge and bulbous nasal tip, high-arched palate; a short and broad neck and mild clinodactyly of the fifth fingers. (Fig. 1) On examination of the genitalia absence of the scrotum and a small penis were noted. (Fig. 2) Testes were not palpable in the inguinal channels.

The boy could not walk without support. Speech was absent, but he responded to commands given by the examiner, and he was able to point to objects on request.
Neuropsychiatric evaluation revealed: moderate mental retardation (I.Q. = 54), severe deficiency of expressive language and spastic paraparesis.

X-ray examination of the knees showed bilateral absence of patella. (Fig. 3) MRI investigation of the skull did not reveal any abnormality of the skull or of the brain. (Fig. 4) MRI of the pelvis demonstrated the absence of testes in the inguinal channels, and did not reveal their presence in the pelvis either. (Fig. 5)
facial anomalies described in duplications of 1p are inconsistent; some of the features of our patient have been noted before: short palpebral fissures, wide nasal bridge and bulbous nasal tip, bilateral ptosis,6,8 Clinodactyly of the fifth fingers has also been reported.5,7 So far, the absence of patellae has been a unique finding.

Two cases having the duplication 1p have been reported: a girl with a de novo presumed inverted duplication of (1)(p11-p31) and a boy with a de novo presumed direct duplication of (1)(p21.2-p32).6,8 In neither of these two cases FISH (Fluorescence In Situ Hybridization) was performed to confirm that the duplication segment was from chromosome 1. We found only one such case in the literature which was confirmed by FISH analysis after it was identified by GTG banding.10

CONCLUSIONS

We have described a patient with partial duplication of 1p, who presents most of the common clinical features described in cases with similar duplications, except growth retardation and brachydactyly. The absence of patellae is a finding that has not been reported before.

The presence of genital abnormalities in our case, just like in all male cases with duplications involving the 1p31.1 region, provides another argument that this region contains a gene with an important role in the development of male genitalia.

REFERENCES


DISCUSSIONS

Up to now, there have been 10 cases reported with partial duplication of 1p, having different breakpoints and size. These cases show a broad range of clinical manifestations, the clinical picture varying from severe (death in the first six weeks of life) to mild-to-moderate mental retardation associated with minor anomalies.1,6 Most of the cases shared low birth weight, microcephaly, cleft or high-arched palate and brachydactyly (the latter being absent in our case). All male patients with duplications including 1p31.1 had cryptorchidism.1,2,7,8 A frequent finding was growth retardation, absent in the case of our patient.