SHORT COMMUNICATION

DE NOVO DUPLICATION OF CHROMOSOME 7 (q21.1-q32); CASE REPORT AND REVIEW OF THE LITERATURE

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ABSTRACT

Cytogenetic analysis of a 1-year-old boy with multiple congenital anomalies revealed partial duplication of the chromosome 7q21.2-q32 band region. His main features included: frontal bossing, small jaw, low-set ears, deep-set eyes, strabismus, drooping left upper eyelid, widely-spaced eyes, short nose, long philtrum, down-curved upper lip, camptodactyly and hypotonia.

Key words: Partial duplication of chromosome 7q

INTRODUCTION

Partial trisomy/duplication of chromosome 7q is associated with a characteristic syndrome of frontal bossing, retrognathia, small jaw, low-set ears, dysplastic ears, deep-set and prominent eyes, strabismus, down-curved upper lip, small mouth, short hands, stiffness of fingers and other joints, joint laxity, scoliosis, reduced muscle tone, hydrocephalus, growth retardation, strabismus, coloboma of iris, drooping upper eyelid, widely-spaced eyes, long eyelashes and short space between eyelids [1-8].

CASE REPORT

A 1-year -old boy was referred to our clinic because of developmental delay. He was the fourth offspring of an unrelated healthy couple and his siblings had no significant health problems. He was born by natural delivery after a full term pregnancy weighing 1,500 gr and head circumstance within the normal range. At examination he had frontal bossing, small jaw, low-set ears, deep-set eyes, strabismus, drooping left upper eyelid, widely-spaced eyes, short nose, long philtrum, down-curved upper lip, camptodactyly, hypotonia, and macrocephaly (Figure 1). He had severe psychomotor retardation, failure to thrive, and poor interaction with the environment.

Cytogenetic analysis of phytohemagglutinin (PHA) stimulated peripheral blood sample was performed and showed additional material on the long arm of chromosome 7 (Figure 2). Chromosomal analysis of both parents revealed normal karyotypes. High resolution chromosomal analysis of the proband showed 46,XY,dup 7(q21.2-q32). Twelve patients with duplication/trisomy of 7q have been reported [1-8]. A summary of clinical findings of patients with partial duplication of 7q is given in Table 1.

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Figure 1. Photograph showing the patient at 12 months of age.

Our patient confirms that partial trisomy/duplication of 7q is associated with macrocephaly, frontal bossing, failure to thrive, psychomotor delay and malformed ears. However, correlation of phenotype-genotype is still problematic, possibly because of the existence of an undetectable cell line in patients with pure partial 7q trisomy or monosomy to explain the great clinical variability between reported patients as has been emphasized by Morales *et al* [9].

To the best of our knowledge, our patient is the first reported case of a duplicated 7q for the region between 7q21.2 and 7q32. We suggest that the 7q21.2 region may be critical for the macrocephaly with frontal bossing in our patient and in the patient of Lukusa *et al.* [4].

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Table.1. A summary of clinical findings of patients with partial duplication of 7q.

Region	Clinical Findings	Reference
7q21.2-q22.1	Mental retardation; macrocephaly; frontal bossing; hyperterlorism; small palpebral fissures with downward slant; lobulated tongue; multiple intrabuccal frenula; oligdontia and enamel hypoplasia; cutaneous syndactlyly of fingers 2,3 and 3,4; broad and short fingertips and fetal pads; broad thumbs; halluces	3
7q21qter	Cleft palate; cerebellar hypoplasia and anomalies of the pancreas, gall bladder and appendix	7
7q22.1q31.2	Mental retardation; strabismus; low-set ears; depressed nasal bridge; small nose; hypotonia	5
7q31.3qter	Skeletal abnormalities; facial dysmorphism; dilated cerebral ventricles; microretrognathia and short neck	6
7q35-qter	Developmental and mental retardation; abnormal ears; kyphosis	4
7q35qter	Cleft palate; severe hypotonia and joint contractures	8

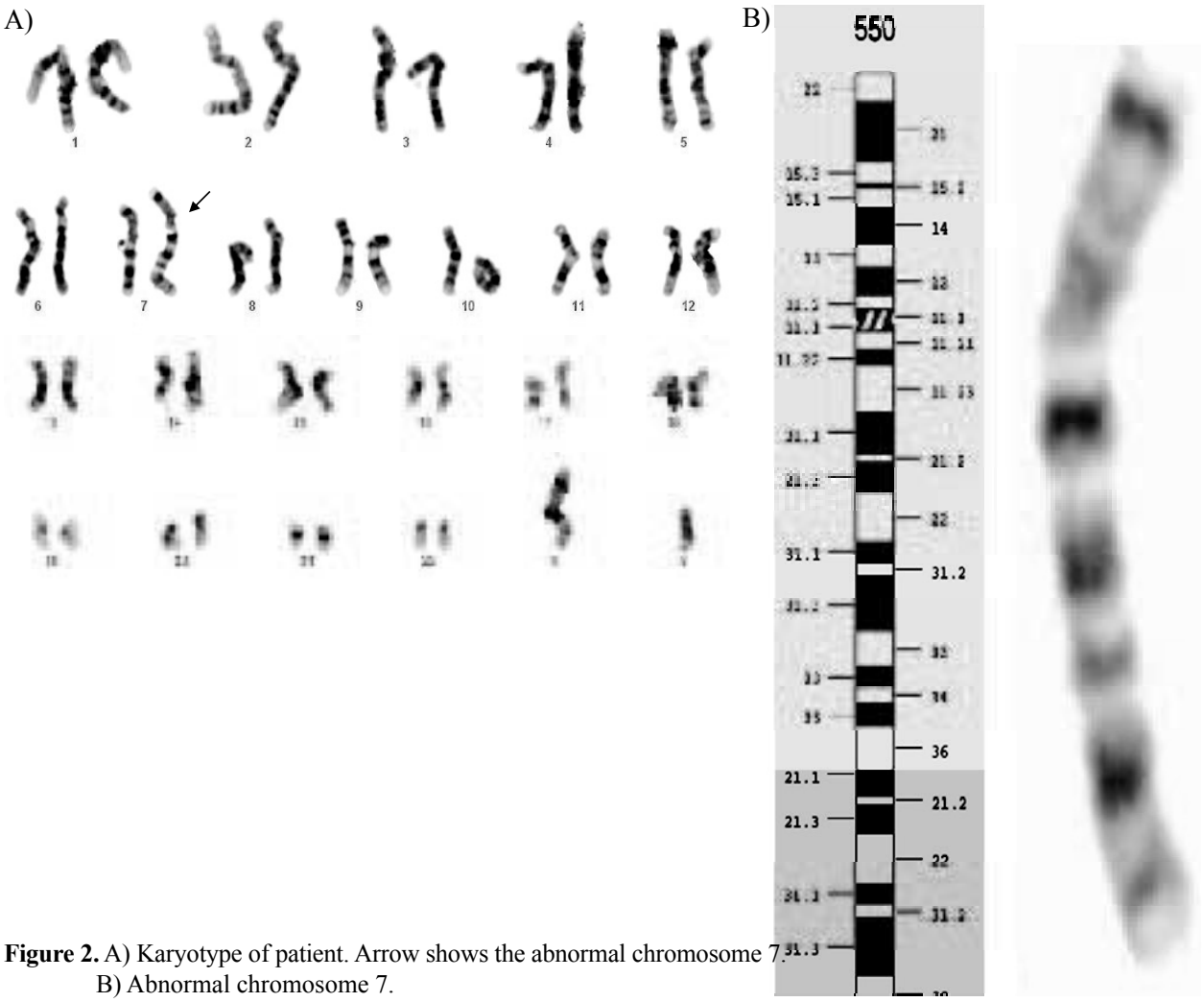

Figure 2. A) Karyotype of patient. Arrow shows the abnormal chromosome 7.
B) Abnormal chromosome 7.

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