

Case Report

Complex Translocation among Chromosomes 2, 3, 9, 15, 18, 20 in a Patient With 3p- Syndrome

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Abstract

A 3-month old girl with monosomy for distal part of the short arm of chromosome 3 is described. Physical examination showed growth retardation, microcephaly, ptosis, micrognathia, low set ears, broad nasal bridge, Simian crease, long philtrum, thin lips and hypertelorism. The patient's clinical phenotype largely resembled that of 3p-syndrome but her karyotype was more complicated than just losing the telomeric portion (3p25.3) of the short arm of one of her chromosomes 3. Her karyotype was 46, XX, t(2;18) (p12;q12.1), del(3) (p23p26), t(3; 9;15;20) (q13;p23;q12;p12). Her parents showed a normal karyotype pattern.

Keywords: Chromosome 3, multiple congenital anomalies, partial monosomy 3p

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Introduction

The 3p deletion syndrome is an uncommon disorder caused by deletions of varying lengths in the 3p25-pter region.

The phenotype of individuals with deletions ranges from normal to severe.¹ Because of its variable expression, it is supposed that this disease has an undefined number of genes that appears to contribute to this contiguous gene syndrome.² Since the first case was demonstrated by Verjaal and De Nef (1978), approximately 30 patients with distal 3p deletions have been described. Despite the low number of patients, different phenotypes, such as developmental delay, low birth weight and growth retardation, micro- and brachycephaly, ptosis, long philtrum, micrognathia, and low set ears, have been described.³⁻⁵ Most cases are *de novo*, although a few familial cases have been reported, as well.¹ The majority of patients carry a terminal deletion of the short arm of the maternal or paternal copy of chromosome 3.⁶

Case Report

Clinical description

The proposita, a 3-month old girl, was the first child of non-consanguineous healthy Iranian parents. The father and the mother were 24 and 19 years old, respectively. They had no history of any specific illness. Physical examination of the patient showed craniofacial anomalies including growth retardation, microcephaly, ptosis, micrognathia, low set ears, broad nasal bridge, Simian crease and long philtrum (Figure1).

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Cytogenetic analysis

Cytogenetic analysis was carried out on the patient and her parents, according to standard cytogenetic procedures.⁷ A minimum of 50 metaphases were analyzed from each sample, using the Applied Imaging CytoVision Karyotyping System (Santa Clara, CA). Karyotypes were assigned according to the recommendations of the International System of Human Cytogenetic Nomenclature (ICSN) (1995). The GTG banded karyotype of the patient showed complicated chromosomal rearrangements. Her karyotype was 46, XX, t(2;18) (p12;q12.1), del(3) (p23p26), t(3; 9;15;20) (q13; p23;q12; p12), (Figure2). Her parents' karyotypes did not show any microscopic rearrangements. To carry out further examination on the patient, informed written consent was obtained from the patient's parents.

Discussion

A few cases of deletion of the short arm of chromosome 3 (del(3) (p25-pter)) who manifest intrauterine and post-natal growth retardation have been reported in the literature. Of these cases, only a few have extra chromosomal abnormalities.^{3,4,8} Fernandez, et al.^{9,10} reported a boy with characteristic physical features of 3p deletion syndrome, together with verbal and non-verbal developmental delay, who carried a *de novo* balanced translocation, t(3;10) (p26; q26). The interesting finding of the case reported here is the extent of rearrangements, which involve chromosomes number 2,3,9,15,18 and 20. The majority of the symptoms manifested by the infant described here are similar to those described in other 3p-syndrome cases,^{6,9,11} except for the Simian crease. Therefore, it is reasonable to assume that the translocation rearrangements detected in our patient are balanced and phenotypically silent, although the level of resolution provided by GTG-banded analysis is not high enough to detect small scale losses of DNA at breakpoints. In this respect, molecular karyotyping could more clearly highlight the chromosome imbalances in our patient, not least characterizing the extent of loss of the terminal end of chromosome 3. Our case appears to be *de novo* as the parents



Figure 1. The proband's facial features at 3 months of age.



Figure 2. Idiogram of GTG-banding of the patient karyotype showed 46, XX, t(2;18) (p12;q12.1), del(3) (p23p26), t(3;20;9;15) (q13.1p12;p23;q12) with multiple chromosomal anomalies.

have normal karyotype. In addition, there is no family history of any significant disorder or documented exposure to any known teratogenic agents during gestation. Future pregnancies could be examined cytogenetically, but the recurrence risk would appear to be low. Finally, given that variable penetrance is associated with 3p- syndrome, a confident prognosis regarding a phenotypic outcome remains challenging.

Consent

The patient has provided written consent for the case report to be published.

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Conflict of Interest

None declared.

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