Case Report

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A CASE OF DELETION OF CHROMOSOME 14q23.3

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SUMMARY

An interstitial deletion of the region q23.3 of chromosome 14 is described in a young male patient with epilepsy, mental retardation, behavioural disorders and severe impairments in social interaction and communication. Interstitial deletions of the long arm of chromosome 14 are extremely rare. However, this case seems to confirm the possible role of genes located on the long arm of chromosome 14 in the maturation of cognitive development and relational abilities.

Introduction

Deletions of chromosome 14 are very rare and have a very large clinical spectrum[1-3]. They may involve an interstitial deletion with variable breakpoints and variable sizes or an apparently terminal deletion of variable size. The deleted chromosome 14 may also be present as a ring chromosome.

We present the case of a young male patient with epilepsy, mental retardation, behavioural disorders and severe impairments in social interaction and communication. These clinical features are related to a microdeletion on chromosome 14q23.3 of 330 Kb.

Case Report

Our proband is a 17 year-old boy, born at term by caesarian section due to fetal malpresentation after an uncomplicated pregnancy. Birth weight was 2650 g. Perinatal distress was present (Apgar score was 1': 3 e 5': 6). The family history is positive for cognitive retardation (both parents and brother). The neonatal period was characterized by hypotonia and torpor. The child had frequent episodes of gastroesophageal reflux and retarded growth. He also showed delay in mental development. He began walking at 18 months, he was clumsy and unable to make precise movements with his hands. His playing was immature with a predominance of sensory-motor play and absence of symbolic-representation play. He had abnormal and impaired development in social interaction and communication (absence of expressive language and eye contact) with presence of motor stereotypies. At the age of three years generalized epileptic seizures occurred and he began pharmacological therapy with lamotrigine and topiramate, achieving a seizure-free state.

His phenotype is characterized by plagiocephaly, facial dysmorfisms and macroglossia. He is affected by eating disorders (hyperphagia) and obesity. Our diagnostic assessment included laboratory tests (blood count, liver and kidney function, muscle enzymes, thyroid function, metabolic screening), genetic testing (karyotype, fragile X, Prader-Willi S.),

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neurophysiological investigations (electroencephalogram), audiological and visual tests, cardiological assessment, neuroimaging (brain CT and MRI). All these investigations were normal.

Discussion

Diagnosis was initially directed toward issues related to perinatal distress. Nevertheless, the idea of a genetic cause is supported by the presence of dysmorphic features, family history of cognitive retardation, severe impairment of communication and relational abilities without deficits of motor functions. This finding was later confirmed by the results of the performance of the Array-CGH genetic technique which revealed a microdeletion on chromosome 14q23.3 expanded 330 Kb. This mutation is not yet described in the literature. However, deletions of chromosome 14 are reported in the literature and they are often characterized by central nervous system involvement in association with the alteration of other systems.

Autism and spherocytosis have been described in a patient with microdeletion of chromosome 14q23.2-23.3 [4]. An interstitial deletion of chromosome 14 involving the region q22.1-q22.3 has been found in a child with bilateral anophthalmia, dysmorphic features, mental delay, language deficit and primary hypothyroidism [5].

Language development disorder and mild cognitive impairment have been shown in a case of deletion on chromosome 14q24-q32.1[6]. Autism, petit mal seizures, microcephaly, dysmorphic features and mental retardation have been reported in a female with a deletion of chromosome 14q32.3 [7]. Mental retardation and somatic dysmorphisms have been seen in other patients with a deletion of chromosome 14 involving 14q24-q32.1 or 14q31-qter or 14q32.3 [8-10]. This patient who has a microdeletion of chromosome 14q23.3 shows a phenotype related to other genetic aberrations of chromosome 14. The similarity of some clinical features suggests a possible role for genes located on the long arm of chromosome 14 in the maturation of cognitive development and relational abilities.

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