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Case Report

17q21.31 microdeletion syndrome: Description of a case further contributing to the delineation of Koolen-de Vries syndrome

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Abstract

The widespread use of Array Comparative Genomic Hybridization (aCGH) technology has enabled the identification of several syndromes associated with copy number variants (CNVs) including the 17q21.31 microdeletion. The 17q21.31 microdeletion syndrome, also known as Koolen-de Vries syndrome, was first described in 2006 in individuals with intellectual disabilities and organ abnormalities.

We report the clinical, instrumental, cytogenetic and molecular investigations of a boy admitted for epilepsy and intellectual disabilities. We carried out detailed analysis of the clinical phenotype of this patient and investigated the genetic basis by using aCGH. We identified a *de novo* microdeletion on chromosome 17q21.31, compatible with Koolen-de Vries syndrome.

Our case shares some of the typical characteristics of the syndrome already described by other authors: delayed psychomotor development, primarily affecting the expressive language, dysmorphic facial features, and epilepsy. However the clinical outcome was not severe as the intellectual disabilities were moderate with good adaptive and functional behaviour. Epilepsy was easily controlled by a single drug, and he never needed surgery for organ abnormalities.
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1. Introduction

The 17q21.31 microdeletion syndrome is a genomic disorder that has been described simultaneously by three groups in 2006, after several single case reports had been reported earlier in intellectual disabled individuals [1].

The common phenotype observed in almost all affected individuals includes developmental delay,

Abbreviations: Array-CGH, Array-Comparative Genomic Hybridization; CNV, copy number variation; EEG, electroencephalography; VPA, valproate; IQ, intelligence quotient; ADHD, attention-deficit hyperactivity disorder

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hypotonia, a friendly/amiable behaviour and mild, and characteristic dysmorphic facial features [2,3]. Other features that usually render the phenotype severe may include cardiac abnormalities (ventriculomegaly: 38%), skeletal abnormalities (scoliosis/kyphosis: 36%), cryptorchidism (78%), kidney/urologic defects (32%). Epilepsy has been described in more than 55% of the cases [1,4].

Here we describe the clinical phenotype including epileptic and neurobehavioural profile of one patient with a 17q21.31 deletion.

2. Clinical report

The patient is a 14-year-old boy, third of three siblings. His parents are non-consanguineous and there is not a significant family history. He was born at 36 weeks of gestation, after an emergency caesarean section for foetal distress. In the neonatal period hypotonia and a ventricular septal defect, with left–right shunt were diagnosed. The cardiac abnormality resolved spontaneously.

Motor developmental milestones were slightly delayed: the patient started crawling at 10 months and walking without support at 17 months. He had marked speech delay: babbling at 20 months and uttering the first words at 36 months and short sentences at 4 years of age. At that time a physical examination reported mild dysmorphic facial features (macroglossia, prominent maxillary and low-set ears) and absent organ abnormalities. Auditory evoked potentials were performed and the result went normal. At the age of 6 years he entered the first grade at school with support for learning disabilities and severe speech dyspraxia: he was noted to have oral motor dyspraxia, difficulties putting syllables together, repeating simple words and using long and complex words whilst having good language understanding skills and good non-verbal communication. Wechsler's scale evidenced a marked lowering of total intelligence quotient scores (IQ: 45). Although in the absence of seizures at this age he underwent a sleep (Fig. 1A) and awake EEG (Fig. 1B) that showed diffuse spikes, polyspikes or spike and wave discharges over the left parietal–temporal regions on a slow and poorly organised background activity.

The patient came to our attention at the age of 11 because of rare afebrile focal seizures (three in a year). All seizures occurred during wakefulness and were characterised by loss of consciousness, asymmetric stiffening and cyanosis. At that time the interictal EEG showed occipital spikes and sharp and waves complexes mainly over the left hemisphere, worsened by sleep (Fig. 1C). He was commenced on antiepileptic treatment with valproate (VPA) at the dose of 1 g/day and he never experienced seizures since.

A clinical examination showed dysmorphic facial features, namely high flat forehead, thick eyebrows,

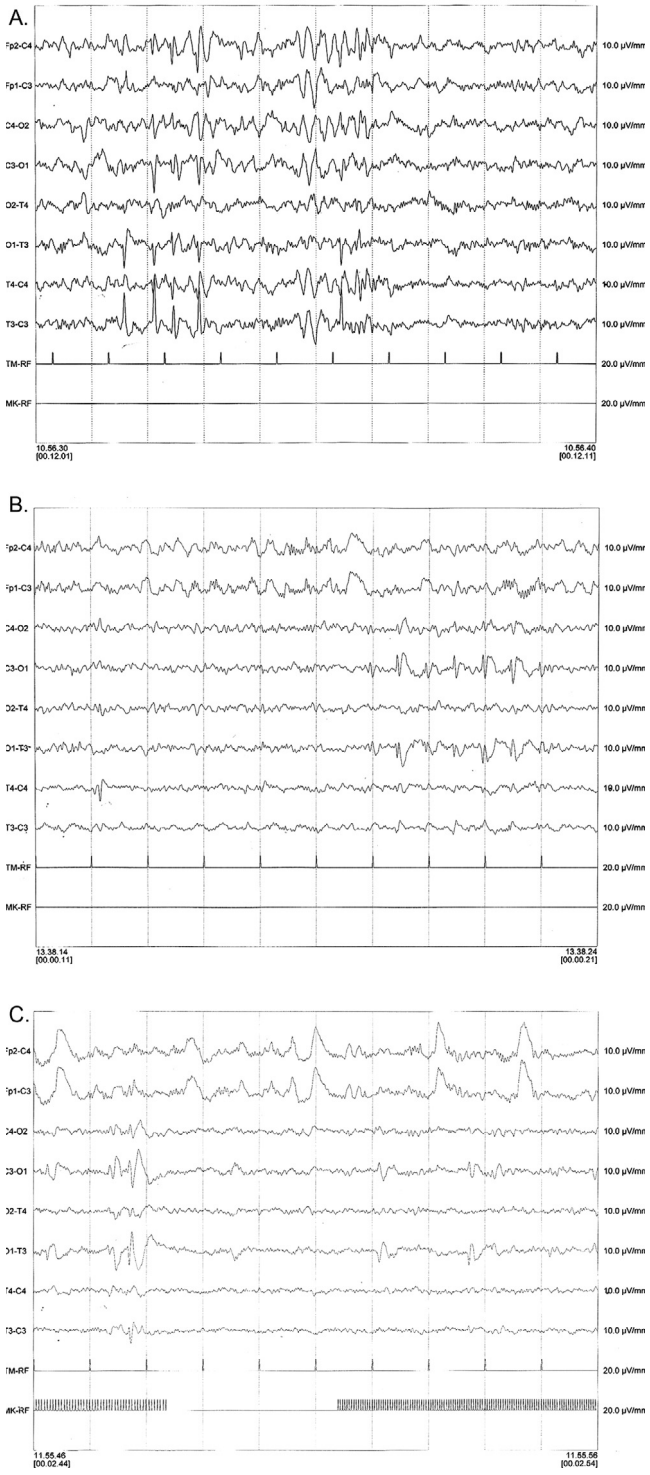

Fig. 1. EEGs of the patients: (A) sleep EEG recorded at the age of six showing diffuse spikes, polyspikes or spike and wave discharges. (B) Awake EEG recorded at the same age showing epileptiform abnormalities, namely sharp and waves complexes over the posterior left hemisphere. (C) Awake EEG recorded at the age of 11 showing spikes and sharp and waves complexes over the occipital region.

macroglossia, prominent maxillary, low-set ears, bulbous nose (Fig. 2A and B). He also showed "elevated laughing and smiling", especially friendly disposition

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