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Review Article

A Comprehensive and Clinical Review of Distal Deletion Syndrome of Chromosome 5q14.3

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Abstract

Cardozo et al. [1] reported 3 unrelated children, 2 boys and 1 girl, with severe mental retardation, epilepsy, and bilateral periventricular heterotopia limited to the subcutaneous region of the temporal bones and occipital lateral ventricles. Using array CGH, Cardoso et al. [1] identified a deletion of chromosome 5q14.3q21 in 3 unrelated patients with periventricular heterotopia.

Keywords: Distal chromosome 5q14.3 deletion syndrome, Chromosomal disorders, Child syndromes

Clinical Signs and Symptoms of Distal Chromosome 5q14.3 Deletion Syndrome

Cardozo et al. [1] reported 3 unrelated children, 2 boys and 1 girl, with severe mental retardation, epilepsy, and bilateral periventricular heterotopia limited to the subcutaneous region of the temporal bones and occipital lateral ventricles. Other features of this syndrome include hypotonia, delayed motor development, lack of speech, and minor facial deformities such as prominent forehead, depressed nasal bridge, and high blood pressure. Also, one of the patients showed polymicrogyria in brain MRI [1] (Figure 1).

Figure 1: Illustration of children with distal deletion syndrome of chromosome 5q14.3 with distinctive facial features [1].

Etiology and Discussion of Distal Deletion Syndrome of Chromosome 5q14.3

Using array CGH, Cardoso et al. [1] identified a deletion of chromosome 5q14.3q21 in 3 unrelated patients with periventricular heterotopia. These deletions ranged in size from 6.3 to 17Mb and included a common region of 5.8Mb. Computational critical region analysis identified 14 candidate genes [1,2] (Figure 2).

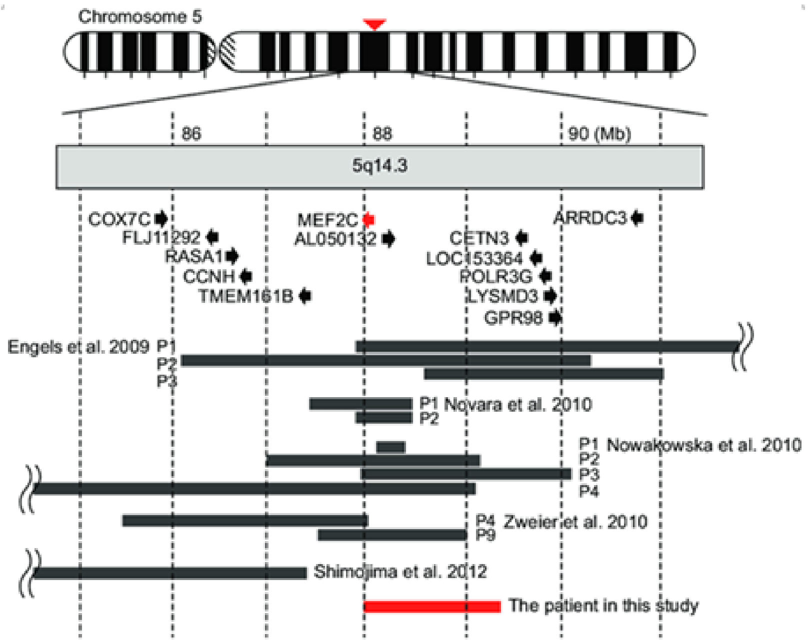

Figure 2: Schematic of the distal deletion mutation of chromosome 5q14.3 [1].

Sobreira et al. [3] identified a 7.4Mb deletion of chromosome 5 at 5q14.3-q21 in an 11-year-old boy with mental retardation, bilateral iris coloboma, hearing loss, dental malformation, and facial deformity. features, but without periventricular heterotopia, which Sobreira et al. [3] referred to the report by Cardoso et al. who identified a deletion region in the overlapping region of 5q14 in patients with periventricular heterotopia. One of these patients had a unilateral coloboma and shared part of the deletion with the patient reported by Sobreira et al. Comparison of the shared deletion regions between the 2 patients revealed a 2.6Mbp putative region for coloboma and a 1.84Mbp putative region for periventricular heterotopia [1,4] (Figure 3).

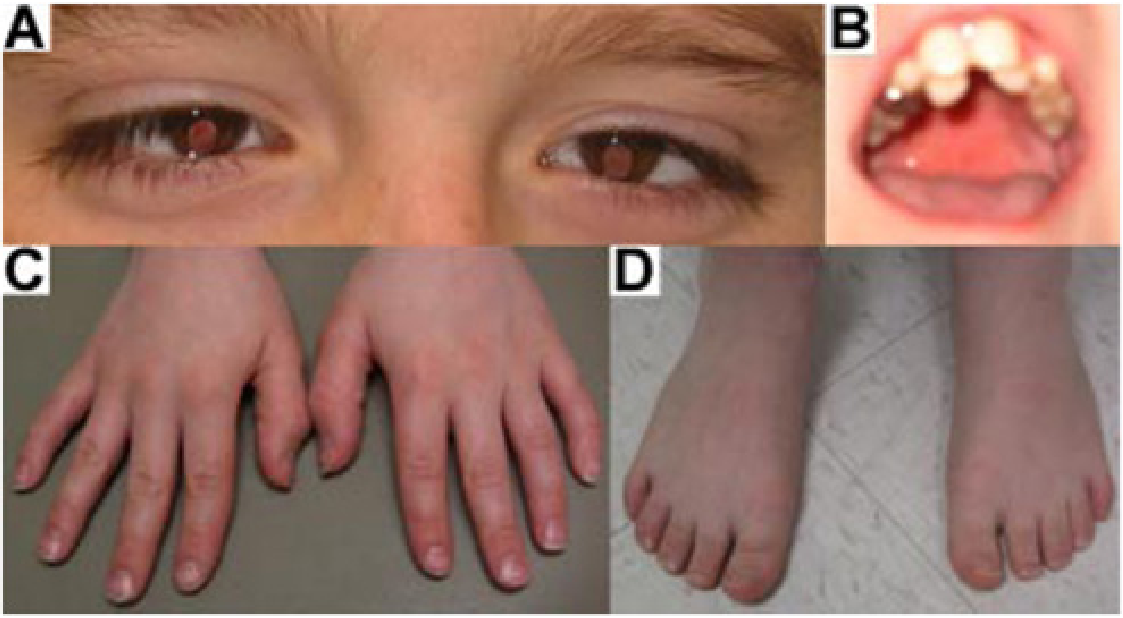

Figure 3: Another view of a child with distal chromosome 5q14.3 deletion syndrome [1].

Le Meur et al. [5] reported 5 unrelated children with severe mental retardation, absent speech and stereotyped movements, each with deletions between different regions of chromosome 5q14 ranging in size from 216kb to 8.8Mb. The minimal common deletion region contained only the MEF2C gene. Le Meur et al. [5] noted that the 5q14 region partially overlapped with that deleted in patients with periventricular heterotopia reported by Cardoso et al. But only one of those patients has deleted the MEF2C gene. Furthermore, none of the patients reported by Le Meur et al. [5] had periventricular heterotopia [1,5].

Al-Kateb et al. reported an 8-year-old boy with a de novo deletion of 582kb on chromosome 5q15, involving 5genes. They compared their patient's findings with those of 3patients reported by Cardoso et al. All of them had a minimum overlapping region of about 230kb including 2genes: FLJ42709 and NR2F1. All 5patients had growth delay and facial deformities, 4 had hypotonia and 3 had eye abnormalities. Urinary tract obstruction was observed only in their patient. Periventricular heterotopia was also present only in the patients reported by Cardoso et al. Al-Kateb, et al. stated that NR2F1 is the strongest candidate gene for overlapping phenotypes. Heterozygous mutations in the NR2F1 gene have been identified in patients with Bosch-Boostra-Schaaf optic atrophy syndrome, which is characterized by developmental delay, moderate mental retardation, and visual atrophy [1,3] (Figure 4).

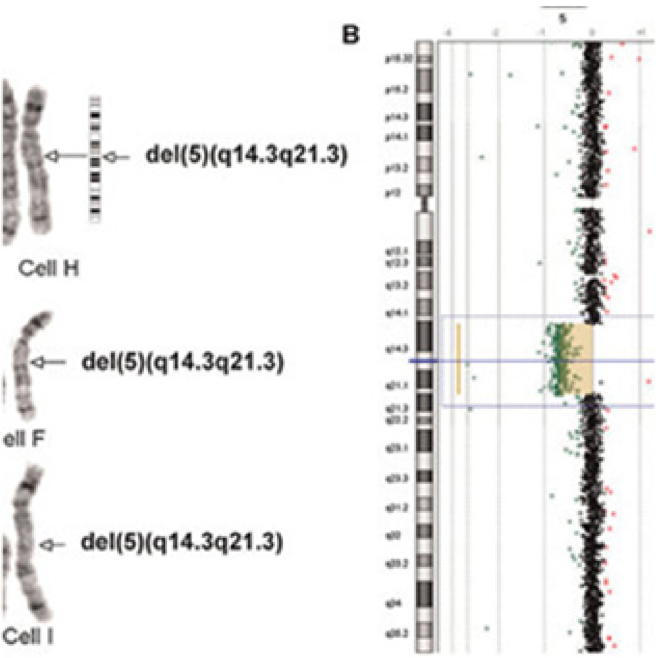

Figure 4: Schematic of the distal deletion mutation in the long arm of chromosome 5 [1].

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