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7p22.1 microduplication syndrome: Refinement of the critical region

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Abstract:
7p22.1 microduplication syndrome is mainly characterized by developmental and speech delay, craniofacial dysmorphisms and skeletal abnormalities. The minimal critical region includes two OMIM genes: *ACTB* and *RNF216*. Here, we report on a girl carrying the smallest 7p22.1 microduplication detected to date, contributing to the delineation of the clinical phenotype of the 7p22.1 duplication syndrome and to the refinement of the minimal critical region. Our patient shares several major features of the 7p22.1 duplication syndrome, including craniofacial dysmorphisms and speech and motor delay, but she also presents with renal anomalies. Based on present and published dup7p22.1 patients we suggest that renal abnormalities might be an additional feature of the 7p22.1 microduplication syndrome. We also pinpoint the *ACTB* gene as the key gene affecting the 7p22.1 duplication syndrome phenotype.

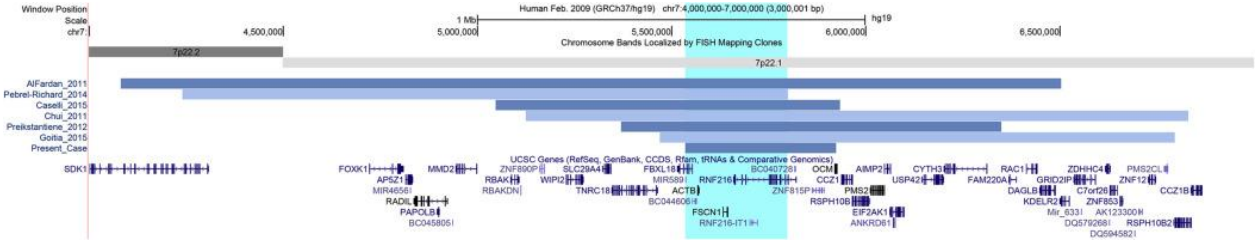

Fig. 2.
Genetic map of the duplicated regions reported in the literature compared with our case. The new smallest region of overlap (chr7:5,536,848–5,799,719) is defined by the distal breakpoint of the duplication of the present case and the proximal breakpoint of the aberration of the Pebrel-Richard ‘s case. The breakpoints are according to the 37 build (Feb 2009) of the Human Genome Reference Consortium (GRch37/hg19).

Fig. 1.
The patient's facial appearance. Note the prominent forehead, high-arched eyebrows, hypertelorism, long and upslanted palpebral fissures, long and prominent philtrum, and microretrognathia.