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5p13 microduplication syndrome: A new case and better clinical definition of the syndrome

Francesca Novara, Enrico Alfei, Stefano D'Arrigo, Chiara Pantaleoni, Silvana Beri, Valentina Achille, Francesca L. Sciacca, Roberto Giorda, Orsetta Zuffardi, Roberto Ciccone

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Abstract:

Chromosome 5p13 duplication syndrome (OMIM #613174), a contiguous gene syndrome involving duplication of several genes on chromosome 5p13 including NIPBL (OMIM 608667), has been described in rare patients with developmental delay and learning disability, behavioral problems and peculiar facial dysmorphisms. 5p13 duplications described so far present with variable sizes, from 0.25 to 13.6 Mb, and contain a variable number of genes. Here we report another patient with 5p13 duplication syndrome including NIPBL gene only. Proband's phenotype overlapped that reported in patients with 5p13 microduplication syndrome and especially that of subjects with smaller duplications. Moreover, we better define genotype-phenotype relationship associated with this duplication and confirmed that NIPBL was likely the major dosage sensitive gene for the 5p13 microduplication phenotype.

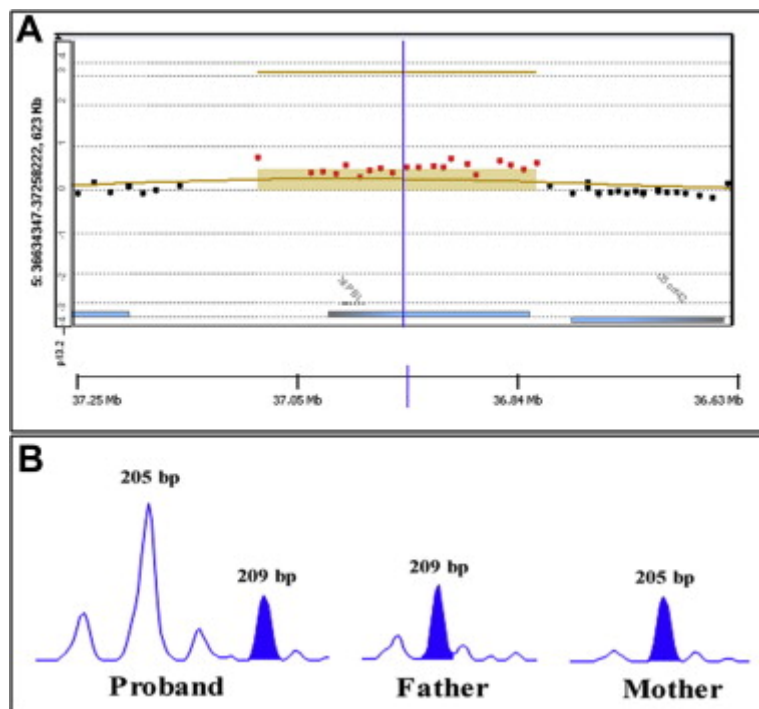


Fig. 1. Array-CGH results: the figure represents the array-CGH profile of our patient. B. Parental origin of the imbalance: genotyping of STS1-chr5p13 microsatellite evidenced the maternal origin of the duplication. Double peak height of the maternal allele in the child indicates that the duplication originated from the maternal chromosome.

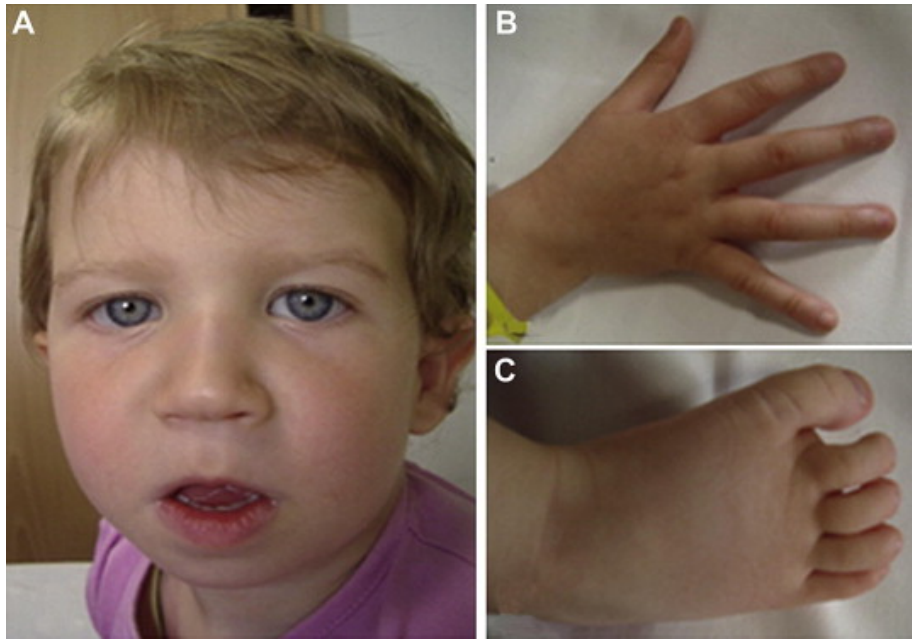


Fig. 2. Photos of our patient showing her dysmorphic features of face and extremities.