



## DE NOVO 17P11.2 MICRODUPLICATION – POTOCKI-LUPSKI SYNDROME DETECTED BY CMA

IRENE PLAZA PINTO; APARECIDO DIVINO DA CRUZ; LYSA BERNARDES  
MINASI; ALDAIRES VIEIRA DE MELO; DAMIANA MÍRIAN DA CRUZ E  
CUNHA; CRISTIANO LUIZ RIBEIRO  
[iplazapinto@gmail.com](mailto:iplazapinto@gmail.com)

**Objetivo:** Potocki-Lupski Syndrome is a continuous gene syndrome caused by the microduplication of 3.7 Mb at 17p11.2 characterized by developmental delay, intellectual disability. Herein, we report a rare case 17p11.2 microduplication detected by Chromosomal Microarray Analysis in Central Brazil. **Método:** Physical examination of the 11 years old boy showed psychomotor development and intellectual disability, and a height of 139.5 cm, a weight of 32.5 g. With Pontifícia Universidade Católica de Goiás ethics approval and informed consent, conventional cytogenetics studies were carried out using peripheral blood, following standardized procedures. The peripheral blood from the proband and his parents was obtained, in order to determine the origin of potential DNA imbalances. Genomic DNA was isolated and the CMA using a GeneChip HD was performed according to array protocols. CEL files obtained by scanning the arrays were analyzed using the ChAS software. Gains and losses with 50 and 25 markers, respectively, in a 100 kb length were considered. **Resultados:** Karyotyping showed a male karyotype (46,XY). CMA detected one genomic imbalance in the patient's genome, corresponding to a de novo 3.68 Mb microduplication at 17p11.2, spanning over described 64 genes. In our study, the use of CMA allowed the identification of microduplication at 17p11.2 in a boy with intellectual disability, reporting the first case of Potocki-Lupski Syndrome in Central Brazil. Several reciprocal deletion/duplication syndrome of chromosome 17 were associated with the dosage sensitive genes. The number of RAI1 copies is most likely responsible for SMS and PTLs. The RAI1 is expressed at high levels mainly in neuronal tissues, and CNV of RAI1 causes distinct neurobehavioral and craniofacial consequences. **Conclusão:** The CMA have made it possible to identify pathogenic CNVs that could explain the molecular etiology of ID. This approached was powerful and efficient method to detect imbalance genomic associated with PTLs that other methods could not identify. Furthermore, it is recommended that the family do the genetic counselling to provide information and help to understand about the syndrome, the familial implications of genetic contribution to disease and the chance of disease recurrence.

**Palavras-chave:** Chromosomal Microarray Analysis. Potocki-Lupski Syndrome. 17p11.2 Microduplicação