

## Abstract 105

### **2q24.2 MICRODELETIONS ENCOMPASSING THE *SLC4A10* GENE ARE ASSOCIATED WITH IDIOPATHIC EPILEPSY AND MENTAL IMPAIRMENT**

Ana Cristina Victorino Krepischi<sup>1,2</sup>, Jeroen Knijnenburg<sup>3</sup>, Debora Romeo Bertola<sup>4</sup>,  
Chong Ae Kim<sup>4</sup>, Fernando Kok<sup>5,6</sup>, Angela Maria Vianna-Morgante<sup>1</sup>, Carla Rosenberg<sup>1</sup>

<sup>1</sup>Department of Genetics and Evolutionary Biology, Institute of Biosciences, University of São Paulo, São Paulo, Brazil; <sup>2</sup>A.C. Camargo Hospital, São Paulo, Brazil; <sup>3</sup> Department of Molecular Cell Biology, Leiden University Medical Center, Leiden, The Netherlands; <sup>4</sup>Clinical Genetics Unit, Instituto da Criança, Hospital das Clínicas, University of São Paulo, Brazil; <sup>5</sup>Department of Neurology, Hospital das Clínicas, University of São Paulo, Brazil; <sup>6</sup>Fleury Medicine and Health, São Paulo, Brazil.

*e-mail:* [ana.krepischi@gmail.com](mailto:ana.krepischi@gmail.com)

Since comparative genome hybridization on microarrays (array-CGH) was introduced in the clinical setting, a high frequency of submicroscopic rearrangements has been detected in cases of idiopathic mental impairment associated with a range of other clinical features. About 10-15% of these patients carry *de novo* microdeletions or duplications likely to be the cause of their abnormal phenotypes. Here, we describe three cases of *de novo* partially overlapping 2q24.1-q24.3 microdeletions identified in syndromic mentally retarded individuals. The patient's phenotypes were quite variable, but mental retardation was a common feature, and epilepsy was present in two of the patients. Previous publications indicate that deletions of a cluster of genes that code for subunits of voltage-gated sodium channel (*SCN*) are likely to be responsible for the frequent seizures in patients with 2q24 deletions. Although the breakpoints were unique for each of the three deletions, there was a ~590 Kb overlapping segment at 2q24.2 comprising 5 genes. These results point to the *SLC4A10* gene (solute carrier family 4, sodium bicarbonate transporter, member 10) as an important candidate gene for epilepsy.