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Determination of *ABC*B1 and *ABCC2* gene variants in drug-resistant epilepsy probands at Van Buren Hospital in Valparaíso, Chile

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Epilepsy affects 1-2% of population worldwide. Despite treatment, about 1/4 of probands develop drug-resistant epilepsy (DRE). Polymorphisms of multidrug pumps in blood brain barrier have been linked to DRE, particularly on *ABC*B1 and *ABCC2* genes. Our aim was to search for association between *ABC*B1 and *ABCC2* polymorphisms and DRE in Chilean probands. Epilepsy probands (n=140), diagnosed with according to ILAE were classified in two groups; those who qualified within DRE diagnosis (two or more trials of adequately chosen and tolerated drugs without seizure freedom within one year) and drug responsive probands. All probands were interviewed to recollect clinical and epidemiological data. Genomic DNA was extracted by standard lysis buffer procedure from saliva samples. Determination of *ABC*B1 C3435T and *ABCC2* c.-24C>T polymorphisms was performed by PCR-RFLP, as previously reported in literature. We successfully replicated the SNP calling methodology described using commercial human DNA panels. Allelic distribution of *ABC*B1 and *ABCC2* polymorphisms do not significantly vary from those reported in literature and UCSC Genome browser. To date, our data indicate that both *ABC*B1 C3435T and *ABCC2* c.-24C>T have similar allelic distribution in Chilean epilepsy probands to those reported in literature. Patient recruitment is ongoing, and determination of SNP frequency is currently underway. We are currently determining putative differences in allele frequencies in drug-resistant vs. responders epilepsy patients.

Biography

Pablo R Moya has completed his PhD from Universidad de Chile and Post-doctoral studies from National Institute of Mental Health, USA. He is Associate Professor at Universidad de Valparaíso, Chile leading the Neurogenetics Lab and Deputy Director of Nucleo Milenio Biología de Enfermedades Neuropsiquiátricas nuMIND, a Center of Research Excellence in Chile. He has published more than 20 papers in reputed journals and has been serving as an Editorial Board Member of repute.

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