POSTER - DNA AND GENOMICS

PUBLIC GWAS DATA UNVEIL POTENTIAL CHILDHOOD ACUTE LYMPHOBLASTIC LEUKEMIA-ASSOCIATED POLYMORPHISMS IN LNCRNA REGIONS FOR FUNCTIONAL STUDIES IN BRAZILIAN POPULATIONS

Marco Antonio Campanário Sampaio (macscampanario@gmail.com)

Ana Laura Bernhard Beal (analaurabbeal@gmail.com)

Wesley Rodrigues Zabot (wezzbot@gmail.com)

Carlos Gabriel Alves De Lima (carlos.gabriel.alves@outlook.com)

Letícia Pontamianos (leticiapontamianos@ufpr.br)

Mara Albonei Dudeque Pianovski (mpianovski@erastogaertner.com.br)

Daniela F. Gradia (danielagradia@gmail.com)

Carolina Mathias (carol.mathias1@hotmail.com)

Jaqueline Carvalho De Oliveira (jaqueline.carvalho@ufpr.br)

Acute lymphoblastic leukemia (ALL) is the most common childhood's cancer type,and is comprised of multiple subtypes with distinct genetic alterations and patient outcomes. While the genetic basis of ALL susceptibility has been supported by several studies, including genome-wide association studies (GWAS), most studies have focused on the coding portion of the human genome, leaving the analysis of non-coding regions, including long non-coding RNAs (lncRNAs), underexplored. LncRNAs have important regulatory roles in

cancer and are closely related to several tumor development hallmarks,, such as genomic instability and proliferative signaling sustaining. Because of that, these molecules represent novel candidates for diagnostic and prognosis biomarkers. Genetic polymorphisms in lncRNA regions have been associated with susceptibility and protection in cancer types, but studies focused on the association of single-nucleotide polymorphisms (SNPs) with lncRNAs in ALL are lacking, mainly in underrepresented populations. To investigate lncRNA-SNPs associated with pediatric ALL risk, we conducted a GWAS data mining analysis using GWAS Catalog database (https://www.ebi.ac.uk/gwas/), selecting potential SNPs candidates for functional studies in a Brazilian cohort from Erastinho Oncopediatric Hospital (Curitiba, Paraná). We found 12 potential ALL-associated SNPs (rs7156960, rs630662, rs10018622, rs11978267, rs11155133, rs7738636, rs75777619, rs28665337, rs4617118, rs2069426, rs9290663, rs10170236) identified in 10 different lncRNAs regions (AC016526.2, AC025508.1, AC079921.1, AC124014.1, AL035446.1, AL355612.1, CCDC26, CDKN2B-AS1, KCNMB2-AS1, MMADHC-DT), some of which are poorly studied while others are characterized by tumor suppressor and cell cycle regulation activities based on public gene ontology (GO) data. We also designed primers for allele-specific PCRs of each candidate SNP to validate their presence in the Brazilian cohort. So far, we have 74 ALL DNA samples and allele-specific optimized PCR for the genotyping of 2 SNPs (rs9290663 and rs2069426). The results from this work may contribute to association studies and functional analysis in childhood leukemia biology, specifically in the Brazilian population.