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Screening of genetic variants in familial case of myeloid neoplasm using exome sequencing

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The introduction of the next-generation sequencing technologies on the research of myeloid neoplasms has provided valuable contributions on the identification of new molecular biomarkers, more accurate risk ratings and more assertive treatments. This study aimed to identify through exome sequencing specific genetic variants in a family with two sisters (39 and 40) with primary myelofibrosis and history of longtime exposure to pesticides DDT-type. To investigate the genetic variants in these patients, we proceeded to the whole-exome sequencing of DNA samples from bone marrow CD34+ hematopoietic stem cells and germline cells from peripheral blood (CD3+ lymphocytes). The samples were sequenced on HiSeq 2500 (Illumina, Inc.), strict mode and the bioinformatics analysis was conducted with the following tools, respectively: NGSQC Toolkit, BWA-backtrack, SAM tools, GATK and ANNOVAR. A set of 110 genes implicated on the pathogenesis of myeloid neoplasms has been selected for the variants filtering. The variants were considered as possibly associated to these neoplasms when they have fulfilled these criteria: Located in exonic regions, coverage values ≥30X and global minor allele frequency <1%, predicted as deleterious by SIFT software and predicted as pathogenic by PolyPhen2 tool. The filtering has identified the *GATA1* Thr263Met in the bone marrow (BM) and peripheral blood (PB) of both patients and the JAK3 Val718Leu in the BM of the younger sister. The change in *GATA*1 is located at highly conserved region of the protein and contains no record in databases of variants to date. Besides, three variants in genes encoding drug-metabolizing enzymes were identified in one or another patient: *CYP3A5 Gly31fs, CYP2A6* Ser467Stop and *CYP2B6* Thr67Met.

Biography

Sara Teresinha Olalla Saad is a Member of the Editorial Board of the Brazilian *Journal of Medical and Biological Research* and Referee for the journals: *International Archives of Internal Medicine* and *Clinical and Laboratory Hematology.* Her research interests include functional investigation and characterization of the involvement of new target genes and new therapies for the treatment of myelodysplasic syndromes and in leukemic lineages, clinical and molecular investigation of chronic anemias, including hereditary anemias such as sickle cell disease, deficiency of pyruvate kinase and glucose-6-phosphate dehydrogenase, spherocytosis and elliptocytosis, among others.

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