Next-generation sequencing: emerging technology in the area of hematology and blood disorder.

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In the modern era, whole world has experienced tremendous boost in the field of molecular diagnosis by use of DNA sequencing technology. The human genome contains more than 3 billion base pairs that contain all the information of about our health and well-being. The first whole genome sequence of human was published successfully before decades [1,2]. It was very expensive and paid billion dollars to complete. The cost paid for, was much more worthy as it providing the first fundamental understanding of the structure and biology of human genome and relation with diseases [3]. Initially NGS technology was generated huge amount data of human variant later it was proved that this NGS technology is much more effective in the diagnosis human diseases by use of bio-informative tools to select pathogenic variants. In present days, sequencing costs have dramatically decline and therefore it is now routinely using for diagnosis of many rare inherited diseases including hematology and blood disorder. Genome-wide sequence analysis is already playing an important role in the hematology field [4]. Numbers of review article has been published on genome sequencing and its impact on hematology, which provide a state-of-the-art snapshot of this rapidly moving field [5]. Many researchers in this modern world did a big job of preparing reader to uncovered variations in the human genome through NGS. This new sequencing technology is going to solve the challenges that researchers in the field of hematology are going forward. Now a day's researcher are making disease specific targeted NGS panel, which is helping more quickly and precise diagnosis of specific disease in the field of hematology. Keeping in mind the growing research in the area of molecular diagnosis, how genome-wide analysis has unlocked new avenues of research, diagnosis, and therapy for benign hematologic disorders [6]. Recent advances in molecular technologies, mainly next-generation sequencing, inspire us to apply these technologies as a first-line approach for the identification of potential mutations and to determine the novel causative genes in patients with blood disorders. Researcher have started preparing targeted NGS panels for diagnosis of hematologic malignancies, Red cell congenital hemolytic anemia for diagnosis of all rare cause of hemolytic anemia which covers around 70-80 genes associated with hemoglobinopathies, which will cover gene related alpha (HBA1/2) and beta (HBB) globin gene locus analysis, HBD (Δ-globin) sequence analysis, gene related to RBC membrane protein disorders, RBC enzymopathies genes, congenital dyserythropoietic anemia (CDA) and the inherited bone marrow failure syndromes (IBMFS) are a group of rare genetic blood disorders in which there is usually some form of aplastic anemia (failure of the bone marrow to produce blood), associated with a family history of the similar disorder. (Includes sequence analysis of C15orf41, CDAN1, GATA1, KIF23, KLF1, SEC23B), Diamond-Blackfan anemia panel and genes related to bone marrow failure syndromes [7].

A panel of genes already identified by WES and association studies as responsible of CHA or modulators of the clinical course of the disease is already analyzed by mass sequencing methodology (NGS) including between 60-70 genes. Modifiers, related with sickle-cell formation, vascular adhesion to endothelium, tolerance to hemolysis and acute severe events, and the analysis of the RBC glycolytic enzymes are included. The variants obtained were studied by mapping in the GRCh38/hg38 version of the human reference genome (Alignment: GEM v3.5/Variant Calling: Genome Analysis Tool Kit (GATK) v3.6./Annotation effect: SnpEff v4.1). For the prioritization of variants, filters related to pathogecity and population frequency according to the SnpEff v4.1 and Mutation Taster programs generally used for analysis. Some are previously used clinical databases of hemolytic anemia are Human Gene Mutation Database (HGMD) version Professional, ClinVar, Red Cell Membrane Database Mutations Database (RDMD), Leiden Open Variation Database – PKLR. The allele frequency is generally assessed in the population (1000G and ExAC) and in local database. Finally most of the researchers generally used the in silico predictions of pathogenicity and Sorting Intolerant from Tolerant (SIFT), PolyPhen-2, Mutation Taster, and Mutation Assessor [8,9].

It is an honor for me to serve as the Associate editor of Hematology and blood disorders; I sincerely thank the entire author for their outstanding and overwhelming contributions to the journal. I am grateful the editor in chief, the editorial board and staff of the editorial office, lastly I entreat all our valuable readers to submit their valuable work for publication in Journal of Hematology and Blood Disorder so the journal could serve as the face of Hematology and bring your outstanding work pride to a limelight.

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