Current Synthetic and Systems Biology

Short Communication

Molecular-biology-2018 Comprehensive Next Generation Sequencing Based Genomic Analysis of The HBB Locus

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Abstract

Hereditary diseases are common in Saudi Arabia. Such diseases include Sickle Cell Anemia and Thalassemia. Recent epidemiological studies report an alarming increase in the prevalence of hereditary blood disorders in Saudi Arabia. The majority of these positive results were in carriers of sickle-cell disease and β-thalassemia. Most of these diseases causing variants are located on chromosome 11 (p15.4) in the HBB gene. We aim to tackle the issue of haemoglobinopathy genetic testing by identifying the most frequent and rare pathogenic variant mutations in the Saudi population utilizing next generation sequencing (NGS).To identify the rare thalassemia variants, we have collected blood samples and extracted DNA of 183 transfusion dependent beta thalassemia patients. We conducted TaqMan genotyping tests of 6 frequent variants; NM_000518.4:c.92+1G>A, c.92+5G>C, c.93-21G>A, c.118C>T, c.20A>T and c.27dupG. After that, NGS was performed on samples with negative/heterozygous results for these variants. In addition, some samples were Sangern sequenced to validate NGS results and eliminate any possible false negative outcome. After initially genotyping 183 DNA samples with the 6 most frequent beta thalassemia mutations, 67 of these samples were sequenced using Ion Torrent next generation sequencing and 12 additional common and rare variants were identified, 8 of the 67 sequenced samples did not have any apparent mutation and failed to justify the disease designation. Sanger sequencing of 13 samples was performed for variant confirmation. TaqMan assay genotyping is a fast, reliable and cost effective technique. Furthermore, TaqMan genotyping followed by NGS has successfully identified and characterized both common and rare pathogenic variations found in HBB. However, further studies are necessary to develop a unified approach towards SCD and β-thalassemia treatment applicable to all patients affected by these disorders utilizing currently available new technologies in gene editing. In spite of the fact that there are in excess of 20 000 qualities in the human genome, the quantity of qualities that are possibly identified with disease was seen as around 500, which incorporates driver genes of malignancy. With this predetermined number of qualities for examination, it is conceivable to complete profound sequencing, which is a technique to build precision by more than once sequencing a similar site. In the USA, administrations for oncogenic board testing have just started by various endeavors and exploration offices, for example, Foundation One by Foundation Medicine Oncomine by ThermoFisher CANCERPLEX by KEW MSK IMPACT by Memorial Sloan Kettering Cancer Center and OmniSeq Advance by Roswell Park Cancer Institute (In Japan, a large number of these genomic tests created in the USA have been presented and are accessible in the clinical exploration setting. Likewise, unique Japanese boards, for example, NCC Oncopanel by Sysmex Todai Onco Panel by Riken beginning and others, have been created. A portion of the board tests may have a favorable position with a capacity to decide tumor transformation trouble which we portray beneath. Regardless, it is important to confirm the exactness of any board test before it is applied in clinical practice. Albeit a portion of the board tests have been endorsed by the US FDA, it will be necessitated that these board tests be affirmed by the Pharmaceuticals and Medical Devices Agency in Japan for use in future clinical practice. Malignancy quality board testing permits us to break down hereditary transformations treatable with molecular targeted medicates, and investigate the chance of expanded command over the treatment of disease types.In spite of the fact that there are in excess of 20 000 qualities in the human genome, the quantity of qualities that are possibly identified with disease was seen as around 500, which incorporates driver genes of malignancy. With this predetermined number of qualities for examination, it is conceivable to complete profound sequencing, which is a technique to build precision by more than once sequencing a similar site. 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by the US FDA, it will be necessitated that these board tests be affirmed by the Pharmaceuticals and Medical Devices Agency in Japan for use in future clinical practice. Malignancy quality board testing permits us to break down hereditary transformations treatable with molecular‐targeted medicates, and investigate the chance of expanded command over the treatment of disease types. In systems including accuracy medication, choice of the most effective treatment depends on distinguishing a subgroup of patients with specific attributes in their genome. Notwithstanding lung adenocarcinoma, another effective case of a treatment procedure including exactness medication is anti‐human epidermal development factor receptor 2 (HER2) treatment utilizing trastuzumab and other molecular‐targeting drugs for bosom malignancy patients with tumors overexpressing HER2. Bosom malignancy patients with HER2 overexpression at first indicated one of the most exceedingly terrible clinical results because of the organically forceful conduct of HER2‐positive bosom disease cells. By and by, trastuzumab and other recently created drugs that target cells overexpressing the HER2 receptor demonstrated a positive impact on HER2‐positive bosom malignant growth cells and essentially improved patient endurance. Of note, HER2 overexpression has been found in bosom malignant growth, yet additionally in other strong tumors, for example, gastric, colorectal, and lung disease. As of late, it has been uncovered that anti‐HER2 treatments are compelling for patients with HER2 overexpression in different malignant growth types. For example, trastuzumab is the suggested first‐line treatment for cutting edge gastric malignant growth in patients with HER2 overexpression. A cross‐sectional treatment applying a molecular‐targeted sedate for every disease, for example, a “HER2‐oma”, joined by a typical malignant growth quality modification, would now be able to be thought of

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