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Bioinformatics Approach to Support Genetic Research in Autism in Mali

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Abstract : Background & Objectives: Human genetic studies can be expensive, even unaffordable, in developing countries, partly due to the sequencing costs. Our aim is to pilot the use of bioinformatics tools to guide scientifically valid, locally relevant, and economically sound autism genetic research in Mali. Methods: The following databases, NCBI, HGMD, and LSDB, were used to identify hot point mutations. Phenotype, transmission pattern, theoretical protein expression in the brain, the impact of the mutation on the 3D structure of the protein) were used to prioritize selected autism genes. We used the protein database, Modeller, and clustal W. Results: We found Mef2c (Gly27Ala/Leu38Gln), Pten (Thr131Ille), Prodh (Leu289Met), Nme1 (Ser120Gly), and Dhcr7 (Pro227Thr/Glu224Lys). These mutations were associated with endonucleases BseRI, NspI, PfrJS2IV, BspGI, BsaBI, and SpoDI, respectively. Gly27Ala/Leu38Gln mutations impacted the 3D structure of the Mef2c protein. Mef2c protein sequences across species showed a high percentage of similarity with a highly conserved MADS domain. Discussion: Mef2c, Pten, Prodh, Nme1, and Dhcr 7 gene mutation frequencies in the Malian population will be very informative. PCR coupled with restriction enzyme digestion can be used to screen the targeted gene mutations. Sanger sequencing will be used for confirmation only. This will cut down considerably the sequencing cost for gene-to-gene mutation screening. The knowledge of the 3D structure and potential impact of the mutations on Mef2c protein informed the protein family and altered function (ex. Leu38Gln). Conclusion & Future Work: Bio-informatics will positively impact autism research in Mali. Our approach can be applied to another neuropsychiatric disorder.

Keywords : bioinformatics, endonucleases, autism, Sanger sequencing, point mutations
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