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In-silico prediction of candidate SNPs in TRIOBP, TMC1& EYA4 genes causing hereditary deafness in three Sudanese patients using next generation sequencing

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Hereditary deafness comprises a large percentage of all causes of hearing loss. Despite the large number of genetic polymorphisms recognized to cause hereditary deafness, a considerable number of patients do not show any of those. To address the genetic heterogeneity of hereditary hearing loss in Sudanese population, we carried out bioinformatics based analysis of SNPs identified previously by Next Generation Whole Genome Sequencing of three Sudanese patients diagnosed clinically with non-syndromic hereditary deafness. We performed *in-silico* prediction of the structural and functional effects of polymorphisms noted in known deafness causing genes using SIFT and PolyPhen v2. We further studied the stability and 3D structure of the mutant proteins using iMutant and CPH model, respectively. We were able to identify a set of novel SNPs in deafness associated genes in each patient. Novel polymorphisms in (TRIOBP, TMC1, EYA4) genes were found to have the highest prediction scores in both SIFT and PolyPhen. Those novel SNPs showed decreased predicted stability as well as change in 3D structure models. We consider these novel polymorphisms as candidates for further large scale studies.

Biography

Yousuf Hasan Bakhit graduated in 2011 with Bachelor of dental surgery (BDS). He joined as teaching assistant at the department of basic medical sciences, faculty of dentistry university of Khartoum since March 2013, faculty of medicine, university of Khartoum. He is also the administrator of Sudanese Parkinson's Disease research project, one of the founders of Sudan Neuroscience Research Groups (SNRG), one of the two founders of Molecular dentistry lab, Faculty of dentistry, university of Khartoum. His current interest is neuronal dynamics and cognitive neurology.

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