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Investigation of Monogenic Obesity in a Qatari Cohort of Severely Obese and Normal-Weight Controls

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Monogenic obesity is a rare form of obesity that results from rare single gene variants. There are at least nine well-established monogenic obesity genes including LEP, LEPR, MC4R, PCSK1, SIM1, POMC, BDNF, NTRK2 and ADY3. In this study, we aim to investigate the prevalence of, and characterize, rare variants in the known monogenic obesity genes in severely obese subjects and normal-weight subjects from Qatar Biobank. Whole genome sequencing data for 250 cases and 250 controls was generated using the Illumina 10X platform, and processed using BWA for alignment, GATK for variant calling and SNP eff for variant annotation. After applying variant prioritization steps, we identified a number of promising coding variants within our genes of interest that merits further investigations. Among these, a novel heterozygous mutation in MC4R that was detected in three cases and none of the controls. The mutation is predicted to be pathogenic based on Insilco analysis. Further functional studies are needed to confirm the pathogenicity of the identified variants.

Biography:

Dr. Mashaël Al-Shafai is an Assistant Professor at the Department of Biomedical Sciences at the College of Health Sciences at Qatar University (QU). She obtained her Bachelor degree with honor in Human Genetics from Leeds University in the United Kingdom in 2009. After that, she joined the Research Division at Qatar Foundation as a Research Analyst and participated in several research projects at Weill Cornell Medicine in Qatar. In 2016, she acquired her PhD in Clinical Medicine Research from Imperial College London where she investigated the genetics and the epigenetics of obesity in Qatari families using whole genome sequencing and genome-wide DNA methylation profiling technologies. Her research interests lie in the area of medical genetics, particularly on obesity and T2D. She has a number of publications in peer-reviewed journals and has won several research grants for her work. Dr Al-Shafai is currently the co-chair of the IRB committee at QU and she is a member in several committees at QU and at the national level, and a member in the Qatar Genome Project Consortium.