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Using genomic technologies to elucidate modifiers in fragile X syndrome

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Trinucleotide repeat expansion disorders are primarily characterized by the expansion of the triplet repeat from a normal to a pathogenic state. Fragile X syndrome (FXS) is caused by the expansion of the CGG repeat in the 5' untranslated region of the fragile X mental retardation 1 (*FMR1*) gene. Allelic variants of *FMR1* include common (6-44 CGG repeats), intermediate (45-54 CGG repeats), premutation (55-199 CGG repeats) and full mutation variants (>200 CGG repeats). Individuals with the full mutation exhibits FXS with associated autistic behaviors, in addition premutation variants also demonstrate associated disorders including Fragile X-associated primary ovarian insufficiency (FXPOI) found in female premutation carriers and Fragile X-associated Tremor ataxia syndrome (FXTAS) found in both male and female premutation carriers. The genetic architecture of the *FMR1* mutation has not been readily explored in diverse populations, thus there is little data available. We summarize the haplotype structure, interspersion pattern and CGG variants within African, African Americans, Asian and European populations. We use several publically available datasets to compare *FMR1* variants across populations. Further genomic characterization of *FMR1* architecture is needed. Characterization would elucidate structural polymorphisms that predispose carriers and non-carriers to the gene specific triplet repeat expansion.

Biography

Emmanuel Peprah is a program specialist who implements and manages various initiatives within the NIH Immediate Office of the Director. Prior to joining the Office of the Director, he was a research fellow at the National Human Genome Research Institute where he managed the Human Heredity and Health in Africa (H3Africa) Initiative. He has published several manuscripts in the fields of human genetics with a focus on Fragile X Syndrome in African populations and serves on editorial boards of several journals. In 2012 and 2013, he received the NIH Director's Leadership Award for his exceptional project management of NIH initiatives.

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