Genomic variations in African-American and white populations

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Deletions, duplications or rearrangements of genomic regions in the human genomes produce differences in gene copy numbers, referred to as copy number variations (CNV). Those variations account for a substantial portion of human genetic diversity, and in a few cases, have been associated with behavioural traits or increased susceptibility to disease. A study published today in the open access journal *BMC Genetics*, describes a CNV map of the African American genome, and compares frequencies of CNVs between African American and white American/European populations.

Joseph P McElroy and colleagues from the Department of Neurology, University of California at San Francisco, recruited African Americans from 28 States and used their genomes to draw CNV comparisons with the White dataset. "To the best of our knowledge, this is the first detailed map of copy number variations in African Americans. Understanding the distributions of CNVs in a population is a first step to addressing their role in disease".

The authors employed an array of over 500,000 sequences whose position in the human genome is already known due to single nucleotide polymorphisms. They first analysed the interaction of 50 blood samples of healthy African American females with this gene chip platform, and then used the results as a reference to assess copy number variation in samples from a further 385 African Americans, and an additional set of samples from 435 White individuals. In total, 1362 CNVs were detected in African Americans and 1972 in the White cohort. Across most of the

genome, the frequency of CNVs did not differ greatly between the two populations. However, there were two duplications, one on chromosome 15, and one on chromosome 17, whose frequency varied markedly between the two groups.

The research team discovered that the duplication in chromosome 17 (region 17q21) is present in 45% of White but only in 8% of African American individuals. Another independent study has implicated the same region in mental retardation caused by a deletion due to duplication. Among the deleted genes, two of them, CRHR1 (corticitropin releasing hormone receptor 1) and MAPT (microtubule-associated protein tau), were previously associated with some neurological disorders. These two genes are not contained within the 17q21 region of CNV duplication, but map very close to it.

According to McElroy, "It would be good to know if the CNV duplication of the region might have an effect on the expression of these genes, which in turn could result in neurological disease. It is also interesting to find out whether the type of mental retardation associated with this locus is more common in Whites than in Africans or African Americans. If this is true, then it might be one of the first reported diseases with differing ethnic frequencies due to CNVs."

More information: Copy number variation in African Americans Joseph P McElroy, Mathew R Nelson, Stacy J Caillier and Jorge R Oksenberg, BMC Genetics (in press) www.biomedcentral.com/bmcgenet/

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