

The Distribution and Most Recent Common Ancestor of the 17q21 Inversion in Humans

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ABSTRACT

The polymorphic inversion on 17q21, sometimes called the microtubular associated protein tau (MAPT) inversion, is an ~900 kb inversion found primarily in Europeans and Southwest Asians. We have identified 21 SNPs that act as markers of the inverted, i.e., H2, haplotype. The inversion is found at the highest frequencies in Southwest Asia and Southern Europe (frequencies of ~30%); elsewhere in Europe, frequencies vary from < 5%, in Finns, to 28%, in Orcadians. The H2 inversion haplotype also occurs at low frequencies in Africa, Central Asia, East Asia, and the Americas, though the East Asian and Amerindian alleles may be due to recent gene flow from Europe. Molecular evolution analyses indicate that the H2 haplotype originally arose in Africa or Southwest Asia. Though the H2 inversion has many fixed differences across the ~900 kb, short tandem repeat polymorphism data indicate a very recent date for the most recent common ancestor, with dates ranging from 13,600 to 108,400 years, depending on assumptions and estimation methods. This estimate range is much more recent than the 3 million year age estimated by Stefansson et al. in 2005.