

Supplemental Data

ADCK3, an Ancestral Kinase, Is Mutated in a Form of Recessive Ataxia Associated with Coenzyme Q₁₀ Deficiency

Clotilde Lagier-Tourenne, Meriem Tazir, Luis Carlos López, Catarina M. Quinzii, Mirna Assoum, Nathalie Drouot, Cleverson Busso, Samira Makri, Lamia Ali-Pacha, Traki Benhassine, Mathieu Anheim, David R. Lynch, Christelle Thibault, Frédéric Plewniak, Laurent Bianchetti, Christine Tranchant, Olivier Poch, Salvatore DiMauro, Jean-Louis Mandel, Mario H. Barros, Michio Hirano, and Michel Koenig

Figure S1. Genotyping Results of the Genome-wide Scan in Family 1



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The graphic interface (HomoSNP software) reveals shared regions of homozygosity in this consanguineous family. Each panel represents the result of one chromosome indicated on the left of the top bar. Chromosome pter to qter orientation is represented from left to right. Position of known recessive-ataxia genes is indicated by vertical lines on the top bar of each panel. The following bars indicate individual results of the children represented on the left. The regions with more than 25 consecutive homozygous SNPs are in dark blue. The four affected siblings share a region of homozygosity by descent on chromosome 1q41-q42. We have verified that the shared region of homozygosity on 3p is due to homozygosity of the mother who has transmitted one haplotype to patients 1 and 3 and another haplotype to patients 2 and 4.

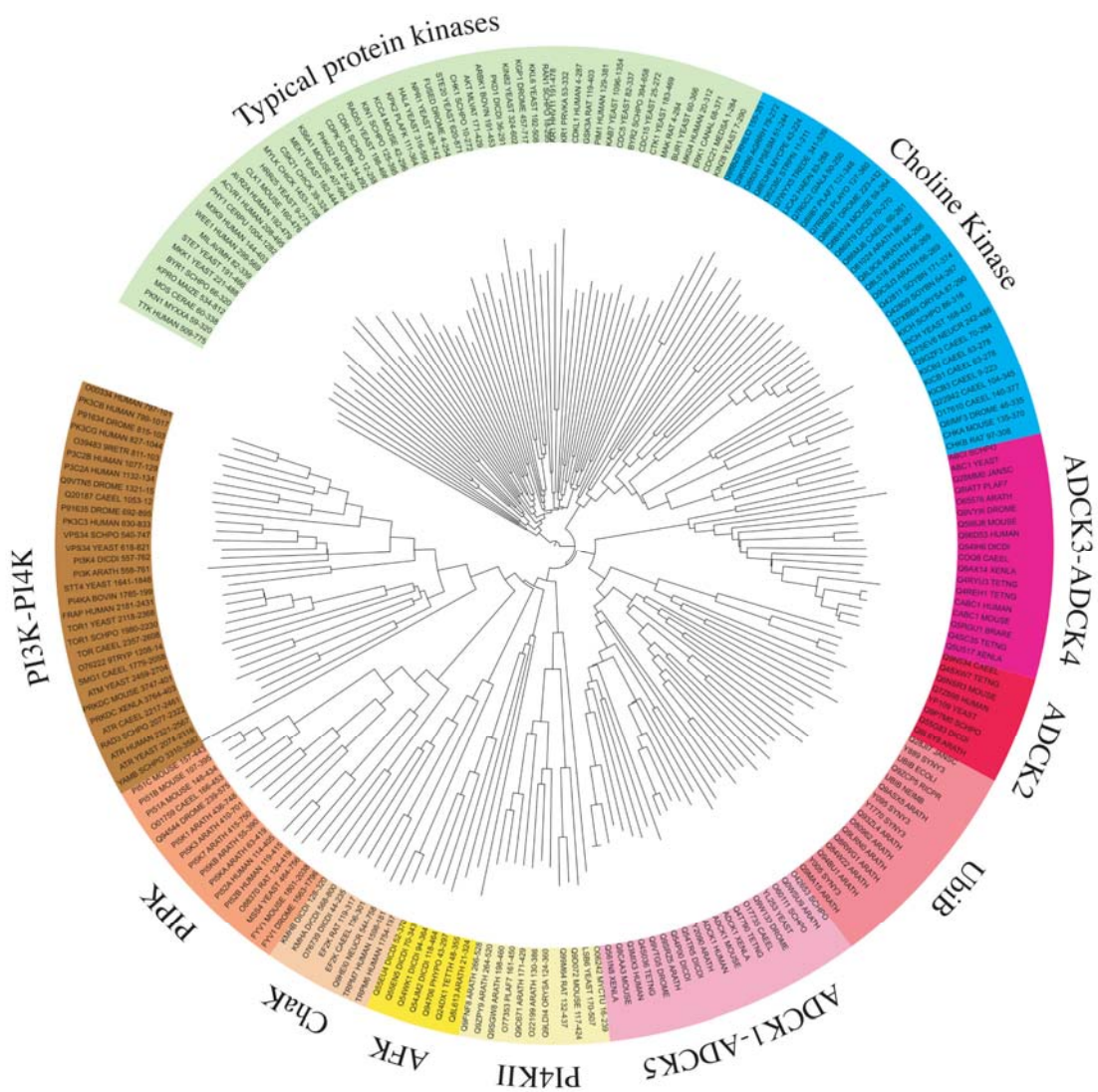


Figure S2. Phylogenetic Tree of Typical and Atypical Protein Kinases

Enlargement of Figure 3B showing SPTREMBL accession numbers. For the non-ADCK proteins, the amino acid position of the kinase “core domain” used for sequence alignment is also indicated. The human ADCKs are as follows: ADCK1 = ADCK1, ADCK2 = Q7Z695, ADCK3 = CABC1, ADCK4 = Q96D53, ADCK5 = Q3MIX3. The *S. cerevisiae* (yeast) ADCKs are as follows: ADCK1-5-like = YL253, ADCK2-like = YP109, ADCK3-4-like = ABC1. The *Jannaschia* sp. (gram-negative bacteria) ADCKs are as follows: ADCK1-5 like = Q28J17, ADCK3-4-like = Q28MM0. Several other alpha-, beta-, and gamma-proteobacteria (gram-negative bacteria) also had one representative of each subgroup (ADCK1-5 and ADCK3-4; not shown).