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Supplemental Data

WDR34 Mutations that Cause Short Rib-Polydactyly Syndrome Type III/Severe Asphyxiating Thoracic Dysplasia Reveal a Role for the NF-κB Pathway in Cilia

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Supplemental Data

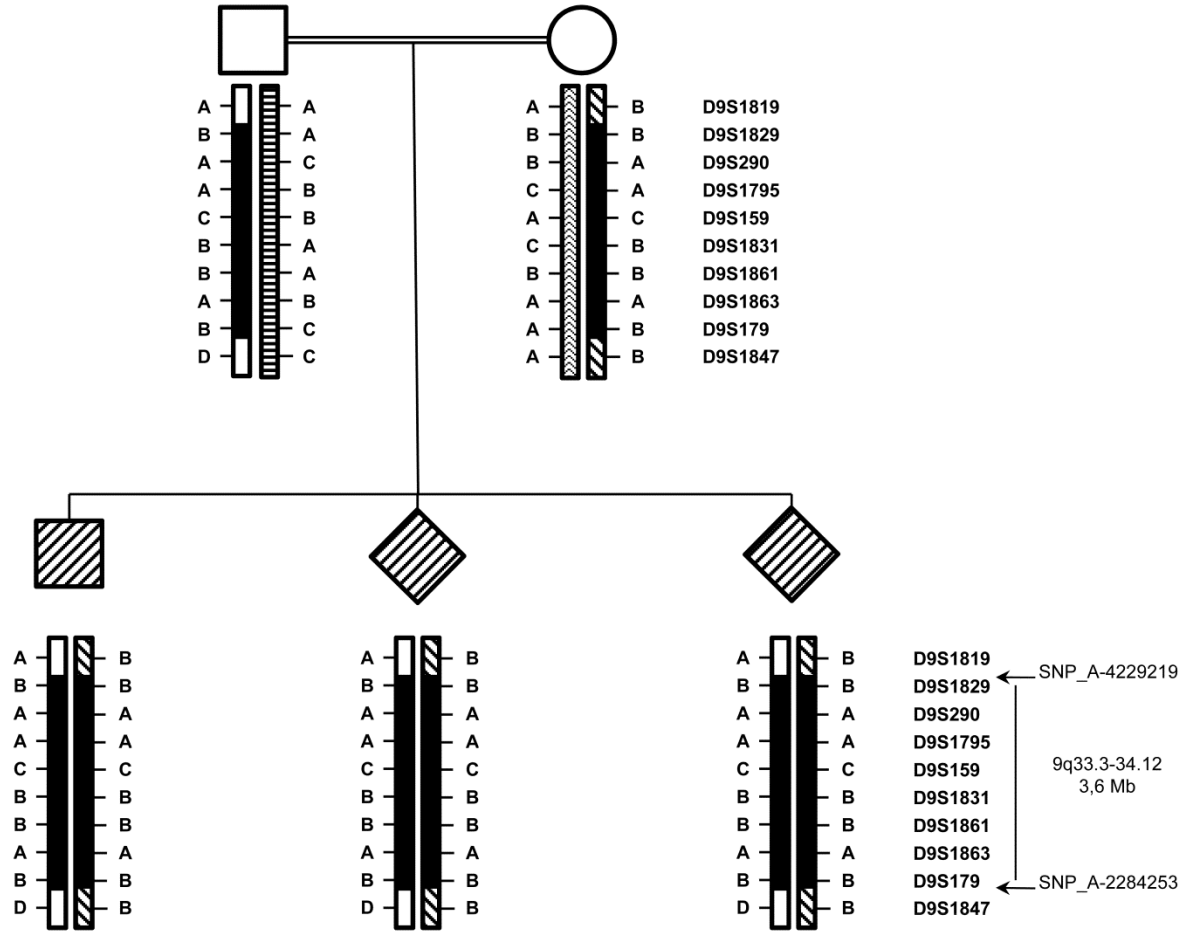

Figure S1. Pedigree and haplotype of family 1 at 9q33.3-34.12

Combining a genome-wide homozygosity mapping (1000 markers, decode genetics) and a GeneChip Human Mapping 250K array, we identified a single region of homozygosity shared by the three individuals on chromosome 9. A critical region of homozygosity of 3.6 Mb was delineated by SNP_A-4229219 and SNP_A-2284253.

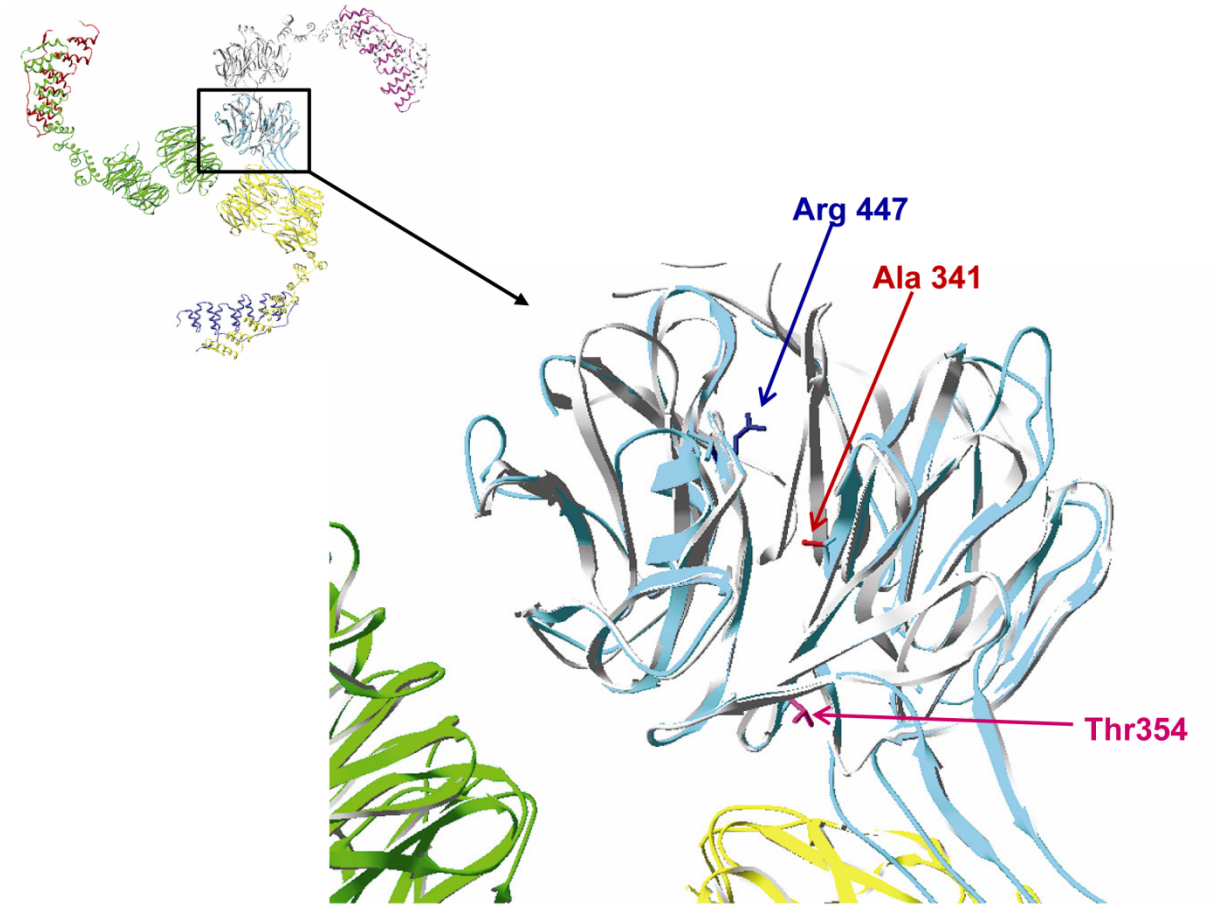

Figure S2. Three-dimensional structural model of WDR34 via Swiss-Pdb Viewer 3.7 representation.

This model highlights the consequences of two of the three mutations: the p.Thr354Met substitution is located in an area that is important for interactions at the triskelion center; the p.Arg447Trp substitution is located in the fourth WD40-repeat (residues 424-464). The functional impact for the p.Ala341Val substitution is not clear from this model.