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Supplemental Data

Mutations in *DDX59* Implicate RNA Helicase in the Pathogenesis of Orofaciodigital Syndrome

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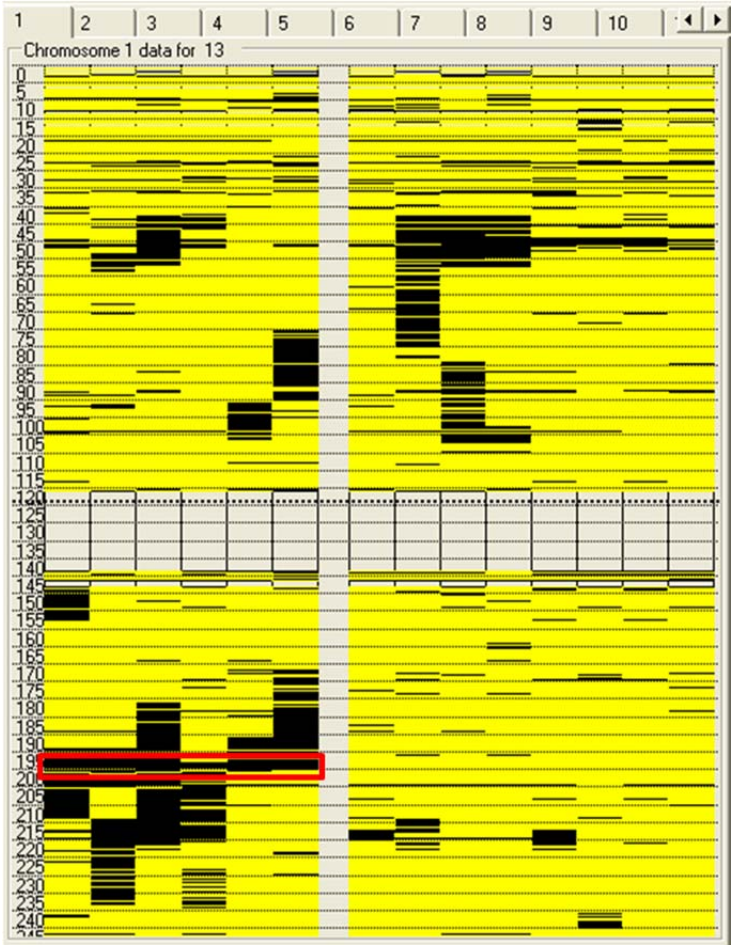

Figure S1. Autozygosity mapping of two families with autosomal recessive OFD. autoSNPa output on the available affected individuals (left) and unaffected relatives (right). Note the exclusive sharing of one black area indicative of homozygosity among affected individuals (boxed in red).

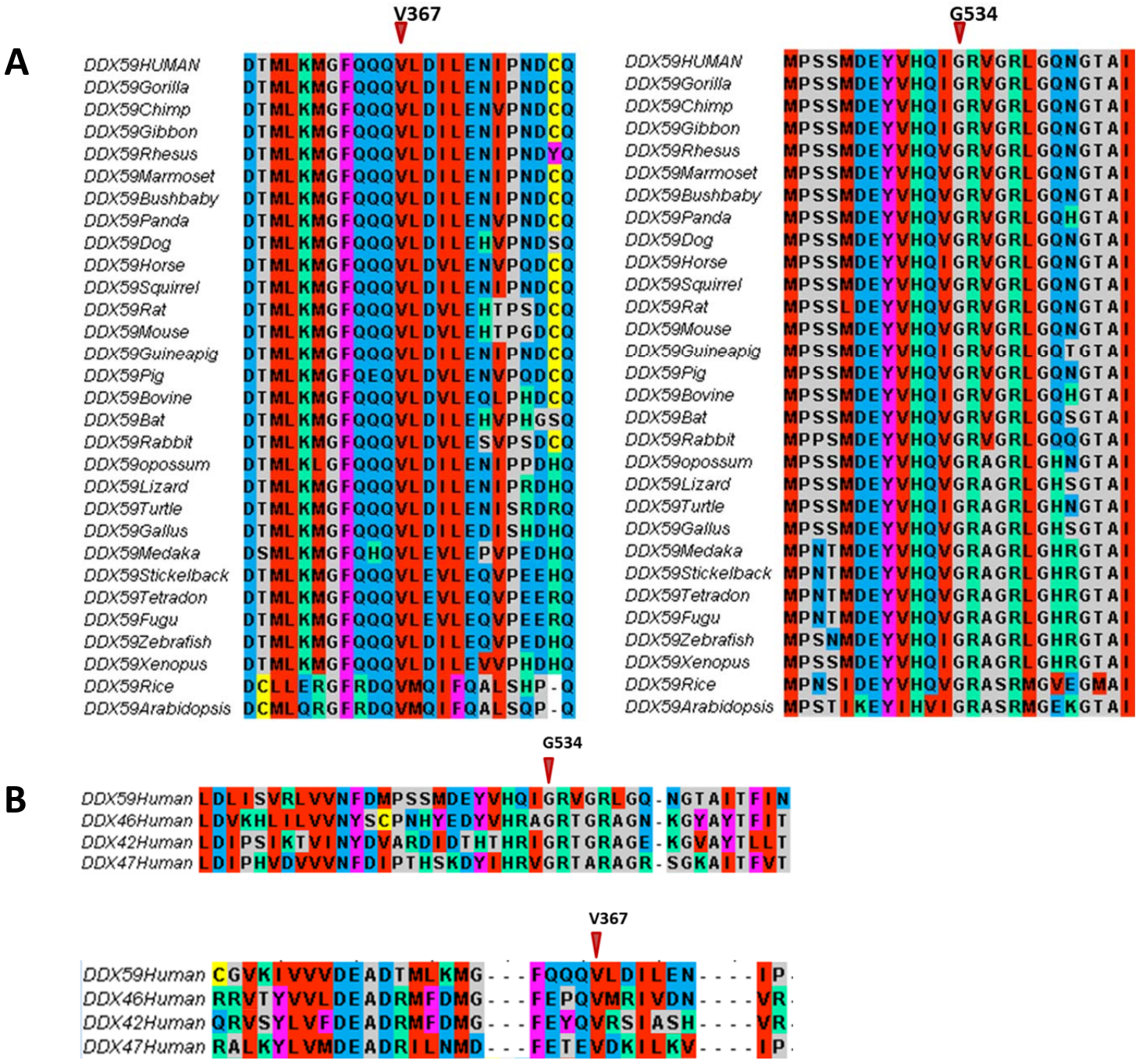

Figure S2. (A) Multispecies alignment of DDX59 showing very strong conservation of the two residues that are replaced by the two mutations reported in the current study. (B) Alignment of DDX59 with other DDX proteins in human showing conservation of the same two residues.

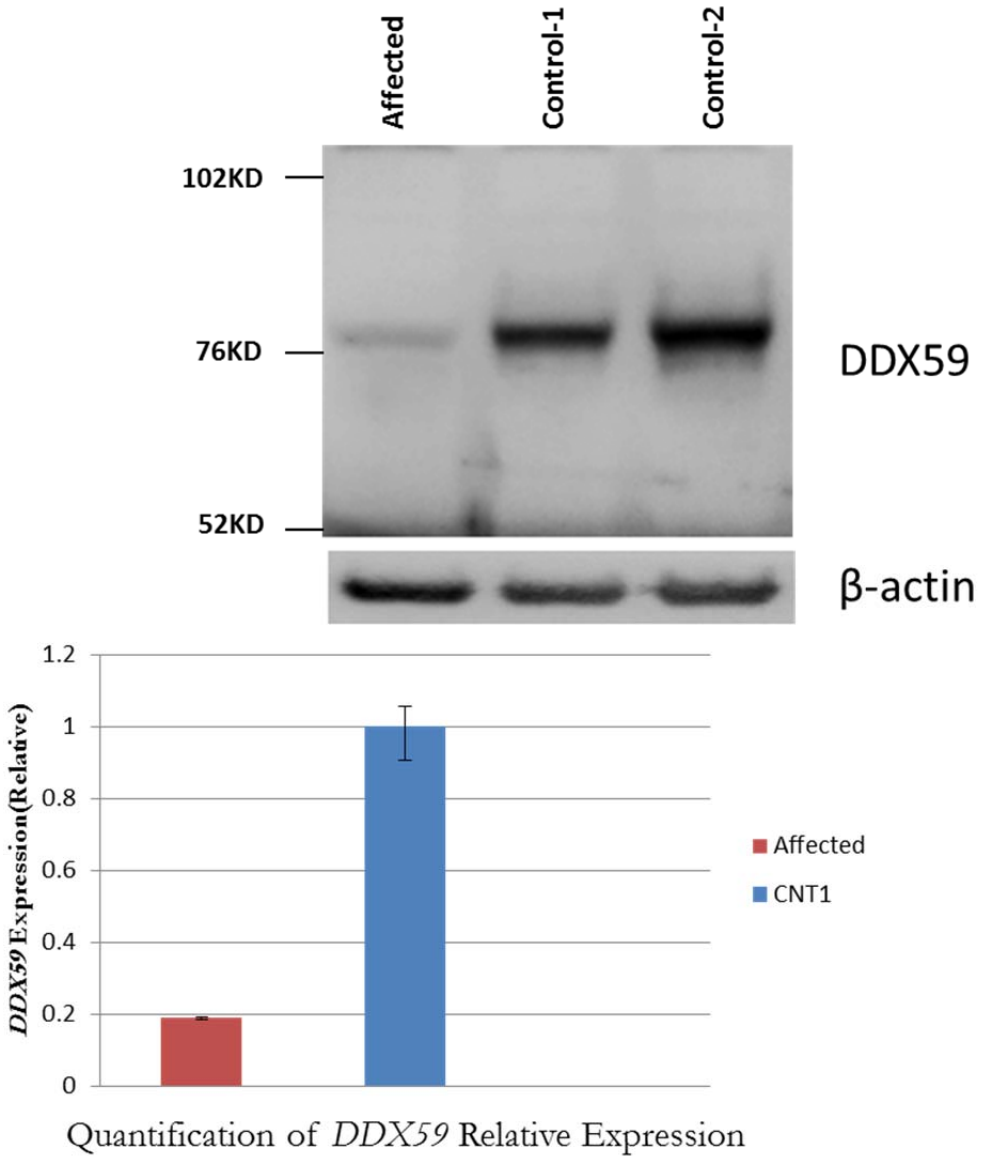

Figure S3. Immunoblot analysis using DDX59 antibody on fibroblasts from two controls and from one affected individual showing marked reduction as quantified in the lower panel.