Additional file 2. **Dias et al. OJRD 2014**

Supplementary figure (.pdf).

Molecular confirmation of Desmosterolosis through exome sequencing and Sanger sequencing

NextGene (Softgenetics, Pennsylvania) view of the exome sequencing reads for one affected child (A) and dideoxy nucleotide sequencing validation (B) for the mother, father, and both affected children. The mother and father were heterozygous for NM_014762.3:c.571G>A (p.E191K) mutation. The propositae have the mutation in homozygosity.