

Supplemental Information

Whole-Genome Analysis Reveals that Mutations in Inositol Polyphosphate Phosphatase-like 1

Cause Opsismodysplasia

Jennifer E. Below, Dawn L. Earl, Kathryn M. Shively, Margaret J. McMillin, Joshua D. Smith, Emily H. Turner, Mark J. Stephan, Lihadh I. Al-Gazali, Jozef L. Hertecant, David Chitayat, Sheila Unger, Daniel H. Cohn, Deborah Krakow, James M. Swanson, Elaine M. Faustman, Jay Shendure, Deborah A. Nickerson, Michael J. Bamshad, and University of Washington Center for Mendelian Genomics

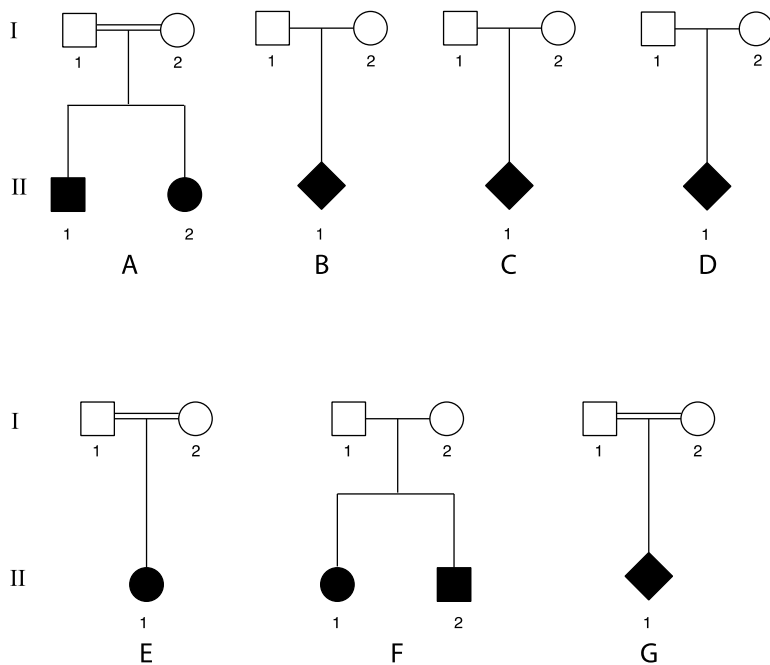


Figure S1. Pedigrees of the seven families in which mutations in *INPPL1* that cause opsismodysplasia were identified.

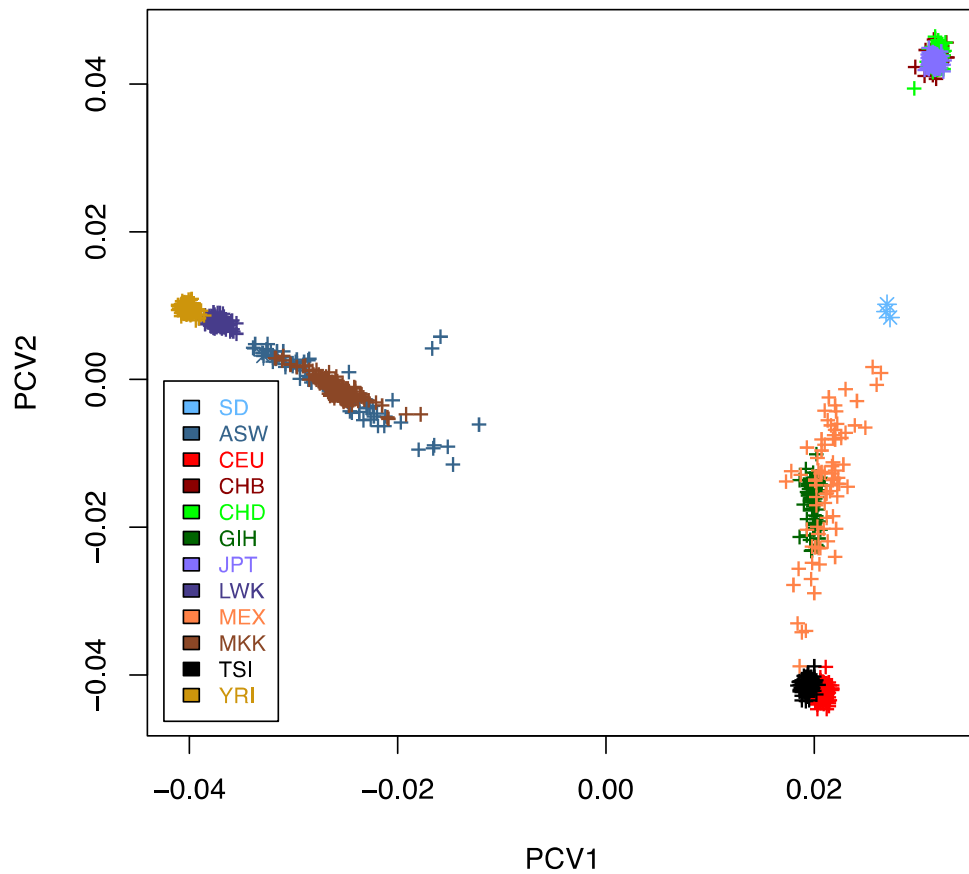


Figure S2. Principal component plot of Family A (light blue, SD – skeletal dysplasia) using genotypes from HumanCytoSNP-12 BeadChips showing clustering is consistent with self-identified ancestry.

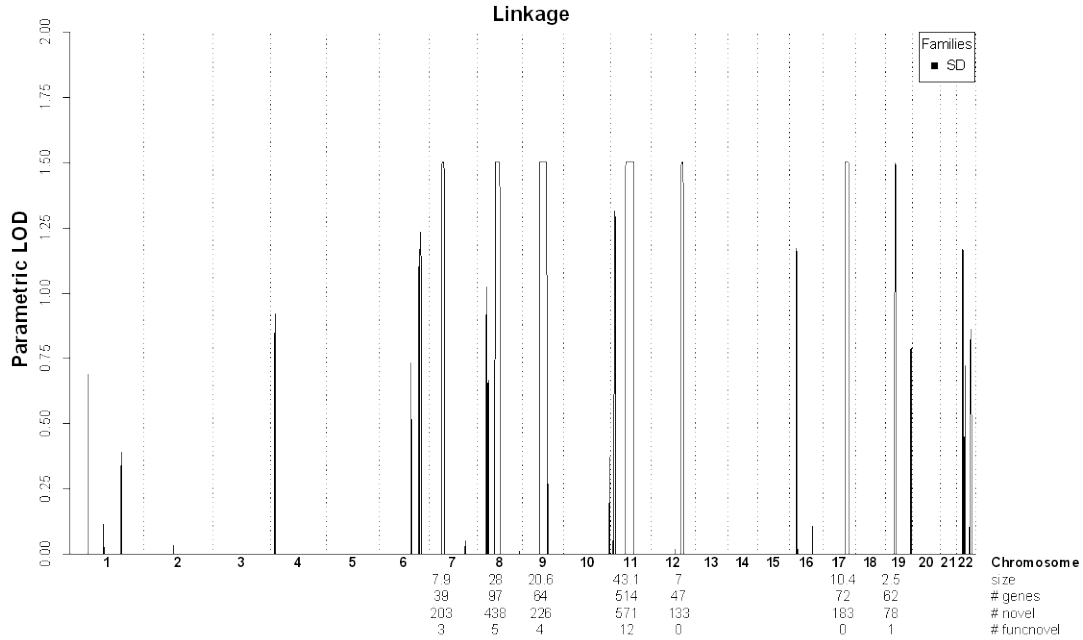


Figure S3. Results of parametric linkage analysis using a fully penetrant rare recessive model ($f_2 = 1$; $q = 0.0001$) and allele frequencies estimated from unrelated members of the HapMap CEPH European, Chinese, Japanese, and Mexican American populations performed using ALLEGRO) on an approximately 0.2 cM SNP map. SD – skeletal dysplasia.