

Supplemental Information

Exome Sequencing Identifies *INPPL1* Mutations

as a Cause of Opsismodyplasia

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Table S1. Filtering Procedure for Bioinformatics Analysis

Individuals	Total Single Nucleotide variations	Variants in neither dbSNP, nor 1 000 genomes nor Exome Variant Server	Variants in neither “In-House” exomes	Variants in neither intergenic, nor intronic, non coding RNA, UTR splicing but essential splicing and Non-Synonymous variants (missense, nonsense, deletion and insertion)	Variants considering homozygote or heterozygote status of patients	Variants considering by SIFT or Polyphen as deleterious/damaging or unknown	Variants considering recessive genetic model
Fam 1 (csg)	67 593	3 860	590	245	19	15	15
Fam 2 (Ncsg)	74 088	4 830	1 945	420	395	148	25
Fam 3 (Ncsg)	69 312	3 656	896	191	179	90	8
Fam 1+2+3	14 870	1 200	54	4	3	2	1