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Supplemental Data

Exome Sequencing Identifies *PDE4D*

Mutations as Another Cause of Acrodysostosis

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Table S1. Filtering Procedure for Bioinformatics Analysis

Number of Individuals	Total SNV	Variant in Neither dbSNP Nor 1,000 Genomes	Neither "In-House" Exome	Nonsynonymous SNP (Missense, Nonsense, Splicing, Deletion, Insertion)	Candidate Gene Involved in cAMP Pathway Signaling
1	46,766	5,431	700	204	2
2	18,083	1,958	334	8	1

Table S2. Genes in Which Novel Variants Were Identified by Exome Sequencing in Two Unrelated Individuals with Acrodysostosis

Genes	Individual 1 Variant	Individual 2 Variant
CD101	p.Leu916Val	p. Tyr61Ser
PEX19	p.Gln82Lys	p.Gln82Lys
GOLGA6B	p.His 618Gln	Splicing
WNK4	p.Ser641Thr	p.Ser1222Arg
FRYL	p.Glu1543Gly	p.Lys1544Glu
PDE4D	p.Pro225Thr	p.Phe226Ser
XPO7	insGG	insGG
MUC5B	p.Ser4378Arg	delC