

Supplementary Table S1. COMP mutations involved in Multiple epiphyseal dysplasia (MED) and in Pseudoachondroplasia (PSACH) according Uniprot databases (UniProtKB - P49747).

Swiss-Prot variant identifier	Position(s)	Amino-acid change	Disorder	References
In EGF-like domain				
VAR_066789	167	G → E	MED	[1]
VAR_066790	234	P → S	PSACH	[1]
In TSP type 3 repeat domain				
VAR_026239	276	P → R	MED	[1], [2]
VAR_066791	290	D → G	PSACH	[1]
VAR_007614	290	D → N	PSACH; mild form	[3]
VAR_066792	298	S → L	MED (overlap with mild PSACH)	[1]
VAR_007615	299	G → R	PSACH	[1], [3]
VAR_066793	311	A → D	MED	[1]
VAR_066794	317	D → G	MED; atypical form	[1]
VAR_066795	326	D → G	MED	[1]
VAR_066796	326	D → Y	PSACH	[1], [3]
VAR_007616	328	C → R	PSACH; mild form	[4], [5]
VAR_066797	341 – 342	Missing	PSACH	[1]
VAR_007617	342	D → Y	MED; Fairbank type	[4], [5]
VAR_066798	348	C → F	MED	[1]
VAR_017102	348	C → R	PSACH	[6]
VAR_007618	349	D → V	PSACH; mild form	
VAR_066799	350 – 372	Missing	PSACH	[1]
VAR_007619	361	D → V	MED; Fairbank type	
VAR_007620	361	D → Y	MED	[5], [7]
VAR_007621	367 – 368	Missing	MED	[5]
VAR_007622	371	C → S	MED; Fairbank type	[1], [8]
VAR_066800	371	C → Y	MED	[1]
VAR_007623	372	Missing	PSACH	[5]
VAR_066801	374	D → N	MED	[1]
VAR_007624	374	Missing	PSACH; mild form	
VAR_066802	376	D → N	MED	[1]
VAR_066803	378	D → V	PSACH	[1]
VAR_066804	385	D → N	MED; atypical form	[1]
VAR_066805	385	D → Y	MED; atypical form	[1]
VAR_066806	385	Missing	MED	[1]
VAR_007625	387	C → G	PSACH; mild form	
VAR_066807	387	C → R	PSACH	[1]
VAR_066808	397	D → H	MED	[1]
VAR_007626	391 – 394	PNSD → V	PSACH	[5]
VAR_066809	402 – 404	GIG → VC	PSACH	[1]
VAR_066810	404	G → R	MED	[1]
VAR_007627	408	D → Y	MED	[5]
VAR_066811	410	C → Y	MED (overlap with mild PSACH)	[1]
VAR_066812	415	N → K	MED	[1]

VAR_026240	420	D → A	MED	[2]
VAR_066813	427	G → E	MED	[1]
VAR_066814	430 – 432	CDS → LWC	MED	[1]
VAR_007628	440	G → E	PSACH; mild form	
VAR_007629	440	G → R	PSACH	[1], [5], [8]
VAR_066815	446	D → N	PSACH	[1]
VAR_066816	448	C → S	PSACH	[1]
VAR_007630	453	N → S	MED; Fairbank type	[9]
VAR_066817	457	Missing	MED	[1]
VAR_007631	459	Missing	PSACH; severe form	[5], [10]
VAR_007632	468	C → Y	PSACH; severe form	[5], [10]
VAR_007633	469	Missing	PSACH; severe form	[5], [7], [11]
VAR_007634	472	D → Y	PSACH; severe form	[5], [10]
VAR_066818	473	D → DD	MED	[1]
VAR_007635	473	D → G	PSACH; severe form	
VAR_066819	473	D → H	PSACH	[1]
VAR_007636	473	Missing	PSACH; severe form.	[1]
VAR_066820	475	D → N	PSACH	[1]
VAR_007637	482	D → G	PSACH	[1], [8]
VAR_066821	501	G → D	MED	[1]
VAR_066822	507	D → G	PSACH	[1], [12]
VAR_066823	511	D → G	PSACH	[1]
VAR_007638	513 – 516	Missing	PSACH; mild form	[8]
VAR_066824	515	D → G	PSACH	[1]
VAR_007639	518	D → N	PSACH; mild form	
VAR_007640	523	N → K	MED; Ribbing type	[1], [13]
VAR_066825	529	T → I	PSACH	[1], [14]
In TSP C term domain				
VAR_007642	585	T → R	MED and PSACH	[1], [9]
VAR_007641	585	T → M	PSACH; mild form and MED1	[1], [2]
VAR_066826	718	R → P	MED	[1]
VAR_066827	718	R → W	MED	[1], [15]
VAR_017103	719	G → D	PSACH; severe	[16]
VAR_066828	719	G → S	PSACH	[1]

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