

# OligoPVP: Phenotype-driven analysis of individual genomic information to prioritize oligogenic disease variants

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**Table 1.** Cases of DIDA combinations missed by OligoPVP prioritization incorporating protein-protein interaction network data, in comparison to ranks of individual variants of each digenic combination ranked by DeepPVP.

DIDA ID	Gene A	Gene B	Disease name (ORPHANET)	DeepPVP Rank A	DeepPVP Rank B	OligoPVP Rank
dd010	NEK1 (C.1640insA)	DYNC2H1 (C.11747G>A)	Short rib-polydactyly syndrome	1	2	Not Found
dd211	MYH7 (C.2645A>G)	RBM20 (C.2062C>T)	Familial isolated dilated cardiomyopathy	2	1	Not Found
dd040	GDAP1 (C.358C>T)	MFN2 (C.479_480delTG)	Charcot-Marie-Tooth disease	1	2	Not Found
dd125	EDA (C.769G>C)	WNT10A (C.511C>T)	Hypodontia	1	2	Not Found
dd218	BMPR2 (C.1471C>T)	KCNA5 (C.1448del)	Heritable pulmonary arterial hypertension	1	2	Not Found
dd244	NLRP3 (C.526C>T)	MEFV (C.442G>C)	Familial Mediterranean fever	2	1	Not Found
dd028	MTF (C.824delA)	TYR (C.1205G>A)	Ocular albinism with congenital sensorineural deafness	1	2	Not Found
dd013	NSMF (C.1132-22.1132-15del)	FGFR1 (C.1025T>C)	Kallmann syndrome	2	1	Not Found
dd018	FGFR1 (C.165_171del)	PROKR2 (C.518T>G)	Kallmann syndrome	1	2	Not Found
dd164	FGFR1 (C.1042G>A)	IL17RD (C.1136A>G)	Kallmann syndrome	1	2	Not Found
dd165	KISS1R (C.581C>A)	IL17RD (C.2204C>T)	Kallmann syndrome	1	2	Not Found
dd166	FGFR1 (C.2075A>G)	DUSP6 (C.545C>T)	Kallmann syndrome	1	2	Not Found
dd168	SPRY4 (C.722C>A)	DUSP6 (C.1037C>T)	Kallmann syndrome	1	2	Not Found
dd169	SPRY4 (C.722C>A)	FGFR1 (C.1447C>A)	Kallmann syndrome	2	1	Not Found
dd117	TYR (C.230G>A)	SLC45A2 (C.1045G>A)	Oculocutaneous albinism	1	2	Not Found
dd121	TYR (C.346C>T)	OCA2 (C.1441G>A)	Oculocutaneous albinism	1	2	Not Found
dd123	LMBRD1 (C.1056delG)	MTR (C.3518C>T)	Homocystinuria without methylmalonic aciduria	2	1	Not Found
dd116	WT1 (C.1228+5G>A)	NPHS1 (C.1126C>G)	Familial idiopathic steroid-resistant nephrotic syndrome	1	2	Not Found
dd050	HMBS (C.422+1G>T)	UROD (C.650_651dupTT)	Porphyria	2	1	Not Found
dd001	KCNQ1 (C.1022C>A)	KCNH2 (C.2592+1G>A)	Familial long QT syndrome	1	2	Not Found
dd048	KCNE2 (C.178T>C)	SCN5A (C.4868G>A)	Familial long QT syndrome	2	1	Not Found
dd065	KCNE1 (C.95G>A)	SCN5A (C.4931G>A)	Familial long QT syndrome	2	1	Not Found
dd069	SCN5A (C.5455G>A)	KCNH2 (C.298C>G)	Familial long QT syndrome	1	2	Not Found
dd134	ATP2B2 (C.1756G>A)	MYO6 (C.737A>G)	Non-syndromic genetic deafness	1	2	Not Found
dd145	KCNJ10 (C.1042C>T)	SLC26A4 (C.919-2A>G)	Non-syndromic genetic deafness	2	1	Not Found
dd206	GJB2 (C.35delG)	TMPRSS3 (C.208delC)	Non-syndromic genetic deafness	2	1	Not Found
dd124	MYO7A (C.2311G>T)	PCDH15 (C.158-1G>A)	Usher syndrome	2	1	Not Found
dd262	TEK (C.309A>C)	CYP1B1 (C.343G>C)	Congenital glaucoma	2	1	Not Found
dd026	MYOC (C.1196G>T)	CYP1B1 (C.1103G>A)	Juvenile glaucoma	2	1	Not Found
dd171	ITGA7 (C.2656G>A)	MYH7B (C.2668C>T)	Left ventricular non-compaction	2	1	Not Found
dd224	TRIM54 (C.316G>A)	TRIM63 (C.739C>T)	Congenital myopathy with protein accumulation	1	2	Not Found
dd019	BAAT (C.226A>G)	TJP2 (C.143T>C)	Familial hypercholelania	1	2	Not Found
dd006	ATP2B2 (C.1756G>A)	CDH23 (C.5663T>C)	Non-syndromic genetic deafness	1	2	Not Found