Table S6: Coding homozygous variants

Patients	Genes	Variant	Human	Mouse
HD09C	*FOCAD*	NC_000009.11:g.[20990242G>A];[20990242G>A]	ND	ND
HK164C	*BRD1*	NC_000022.10:g.[50191485G>A];[50191485G>A]	Associated with schizophrenia	CNS/PNS
	C5orf42	NG_032772.1:g.[84908G>A];[84908G>A]	Joubert syndrome 17 (AR) ALSO HSCR features	Multiorgan abnormalities
	GINS4	NC_000008.10:g.[41399354G>A];[41399354G>A]	ND	ND
	GLRX3	NC_000010.10:g.[131977636A>G];[131977636A>G]	ND	ND
	HK2	NC_000002.11:g.[75105837G>A];[75105837G>A]	Diabetes Mellitus, Insulin-Dependent, 2	Abnormal glucose/insulin metabolism
	ITGB5	NC_000003.11:g.[124578170G>C];[124578170G>C]	ND	ND
	NEK1	NG_027982.1:g.[109891G>A];[109891G>A]	Short-rib thoracic dysplasia. Associated with Polycystic kidney disease (PKD) and amyotrophic lateral sclerosis	Abnormal renal tubule morphology, kidney cysts. Abnormal CNS and craniofacial development.
	PLAT	NG_023264.1:g.[33661T>C];[33661T>C]	Familial thrombophilia	Human tissue plasminogen activator-Cre mouse: aganglionic megacolon
	PPP2R3A	NG_029234.1:g.[142637A>G];[142637A>G]	ND	ND
	RRP7A	NC_000022.10:g.[42912117G>A];[42912117G>A]	ND	ND
	STXBP5L	NC_000003.11:g.[120977910G>A];[120977910G>A]	ND	ND
	USP42	NC_000007.13:g.[6196402A>G];[6196402A>G]	ND	ND
	VRK2	NG_029717.2:g.[256898G>A];[256898G>A]	ND	ND
HK180C	*XRN2*	NC_000020.10:g.[21327108G>A];[21327108G>A]	ND	ND
HK96C	*BICD2*	NG_033908.1:g.[50349C>T];[50349C>T]	Spinal muscular atrophy, lower extremity-predominant 2.	ND
VH105C	*PLEKHA4*	NC_000019.9:g.[49348658C>T];[49348658C>T]	ND	ND
VH108C	*SEMA7A*	NG_011733.1:g.[24026G>A];[24026G>A]	Antigen JMH blood group system. Exotropia. Hemoglobinuria	Olfactory tract is impaired