Table S7: Coding compound heterozygous variants

Patients	*Gene*	Variants (paternal/maternal)	Disorders in humans	Mouse phenotype
HD09C	*BRD4*	NC_000019.9:g.[15391074C>T];[15349596C>A]	Translocation breakpoints in 2 patients with t(15;19)(q13;p13.1) carcinomas	ND
	MAGI3	NC_000001.10:g.[114216074A>G];[114133195T>C]	ND	ND
	FGFRL1	NC_000004.11:g.[1018102G>A];[1018242G>A]	**Wolf–Hirschhorn syndrome (WHS)**	Wolf-Hirschhorn syndrome is recapitulated in Fgfrl1 null mice; Development delay. Many organs affected
	SSH2	NC_000017.10:g.[27958079C>T];[27958200G>A]	ND	ND
	TFR2	NC_000007.13:g.[100238661G>A];[100225717T>C]	Hemochromatosis, Type 3	Abnormal iron homeostasis
HK164C	*DOCK8*	NG_017007.1:g.[176505C>G;253791C>G];[208237C>T]	**Hyper-Ige Recurrent Infection Syndrome, Autosomal Recessive and Autosomal Dominant Non-Syndromic Intellectual Disability**	**DOCK8-deficient mice:impaired induction of antimicrobial peptides in the colon**
	CDC14A	NG_051602.1:g.[159238G>A];[84272C>A]	Nonsyndromic Deafness (AR)	ND
	FRAS1	NG_015812.1:g.[454891A>G];[388680A>C]	Cryptophthalmos, cutaneous syndactyly and genitourinary anomalies (AR)	Abnormal multiorgan development
	SLC24A1	NG_031968.2:g.[39211A>G];[19238G>A]	Congenital stationary night blindness (AR)	Downregulation of Wnt pathway after Wnt3A stimulation
HK180C	*CUL7*	NG_016205.1:g.[21037C>G];[10418C>T]	3M syndrome (AR). Dubowitz's syndrome	Lethal. Respiratory / vascular distress
	ACOX2	NC_000003.11:g.[58510169C>T];[58512313C>T]	ND	ND
	ARFGEF3	NC_000006.11:g.[138531086G>T];[138607972G>C]	ND	ND
	NACAD	NC_000007.13:g.[45121368G>C];[45124560C>T]	ND	ND
HK9C	*LAMA5*	NG_050626.1:g.[53760G>A];[57239G>A]	ND	Arrest of hair development; gloerulopathies; deficient Shh
	RADIL	NC_000007.13:g.[4874645C>T];[4876173A>G]	ND	**Knockdown of radil in zebrafish embryos resulted in multiple defects in neural crest cell-derived lineages including enteric neurons. These defects were primarily due to the diminished migratory capacity of neural crest cells.**
VH105C	*CUL7*	NG_016205.1:g.[12938C>G];[20980G>A]	3M syndrome (AR). Dubowitz's syndrome	Lethal. Respiratory / vascular distress
	ZSWIM4	NC_000019.9:g.[13915803C>T];[13919738G>T]	ND	ND
VH106C	*SYNE1*	NC_000006.11:g.[152730698G>A];[152631870G>A]	SYNE1-Related Autosomal Recessive Cerebellar Ataxia. Emery-Dreifuss muscular dystrophy 4, autosomal dominant (AD)	SYNE2 is required for neuronal nuclear movement and for neuronal migration and development. Hindlimb weakness and an abnormal gait
VH108C	*RFC2*	NG_008102.1:g.[19397G>A];[5089C>T]	Williams-Beuren Syndrome critical region. Infantile hypercalcemia supravalvar aortic stenosis syndrome supravalvar aortic stenosis syndrome	Postnatal growth delay

ND: no described; AR: autosomal recessive; AD: Autosomal dominant; The alleles are ordered by the parental origin: paternal/maternal.