

Supplemental Table 6. Genes possessing disruptive INDEL variants whose mouse orthologs, when disrupted, cause phenotypes.

GENE SYMBOL	No. Of likely disruptive INDEL Alleles	Known disease associations	Mouse Homozygous Disruption Phenotype	Mouse Heterozygous Disruption Phenotype
<i>ABCC1</i>	1	No OMIM	homeostasis/metabolism, cardiovascular system, immune system	Not known
<i>ADAMTS20</i>	1	No OMIM	pigmentation, skin/coat/nails	Not known
<i>ALDH3A1</i>	1	No OMIM	normal	
<i>AP3B2</i>	1	No OMIM	nervous system, behavior/neurological	Not known
<i>ARNT</i>	3	Leukemia, acute myeloblastic	Tumorigenesis, normal, nervous system, renal/urinary system, liver/biliary system, lethality-prenatal/perinatal, homeostasis/metabolism, growth/size, endocrine/exocrine gland, embryogenesis, craniofacial, cellular, cardiovascular system, immune system, reproductive system, skin/coat/nails, hematopoietic system	Normal
<i>ATP8B1</i>	1	Cholestasis, progressive familial intrahepatic 1;Cholestasis, benign recurrent intrahepatic	Liver/biliary system, homeostasis/metabolism, growth/size	Not known
<i>AURKC</i>	1	Male infertility with large-headed, multiflagellar, polyploid spermatozoa	Reproductive system	Not known
<i>BACH1</i>	2	No OMIM	muscle, homeostasis/metabolism, cardiovascular system	Not known
<i>BRWD1</i>	1	No OMIM	reproductive system	Not known
<i>BUB1</i>	1	Colorectal cancer with chromosomal instability	Tumorigenesis, lethality-prenatal/perinatal, homeostasis/metabolism, growth/size, endocrine/exocrine gland, cellular, reproductive system	Tumorigenesis, cellular
<i>C11orf73</i>	1	No OMIM	lethality-prenatal/perinatal	Not known
<i>C16orf35</i>	1	No OMIM	liver/biliary system, lethality-prenatal/perinatal, skin/coat/nails, hematopoietic system	hematopoietic system
<i>C20orf19</i>	1	No OMIM	normal	
<i>CACNA1F</i>	1	Night blindness, congenital	Nervous system, vision/eye	Nervous system, vision/eye.

		stationary, X-linked, type 2A; Cone-rod dystrophy, X-linked; Aland Island eye disease		Human disease model.
<i>CALB1</i>	2	No OMIM	nervous system, behavior/neurological	nervous system, behavior/neurological
<i>CAMKK1</i>	1	No OMIM	behavior/neurological	Not known
<i>CCNB2</i>	1	No OMIM	lethality-prenatal/perinatal, growth/size, immune system, reproductive system, hematopoietic system	Not known
<i>CD207</i>	1	No OMIM	immune system, hematopoietic system	Not known
<i>CEL</i>	2	Maturity-onset diabetes of the young, type VIII	Liver/biliary system, adipose tissue, homeostasis/metabolism, growth/size, digestive/alimentary	Not known
<i>CEP290</i>	1	Joubert syndrome 5; Senior-Loken syndrome 6; Leber congenital amaurosis 10; Meckel syndrome type 4; Bardet-Biedl syndrome 14	Pigmentation, nervous system, vision/eye	Not known
<i>CLCN3</i>	1	Ceroid Lipofuscinosis, Neuronal, 3;	nervous system, liver/biliary system, life span-post-weaning/aging, lethality-postnatal, adipose tissue, homeostasis/metabolism, growth/size, digestive/alimentary, behavior/neurological, immune system, reproductive system, skeleton, vision/eye	Not known
<i>COL4A2</i>	2	No OMIM	lethality-prenatal/perinatal, growth/size, cardiovascular system, embryogenesis, nervous system	Not known
<i>COL7A1</i>	2	Epidermolysis bullosa dystrophica	Normal, limbs/digits/tail, life span-post-weaning/aging, lethality-postnatal, growth/size, digestive/alimentary, skeleton, skin/coat/nails	Not known
<i>CROCC</i>	1	No OMIM	nervous system, immune system, respiratory system, vision/eye	Not known
<i>CSNK2A2</i>	1	Globozoospermia	reproductive system	Not known
<i>CYP27A1</i>	1	Cerebrotendinous xanthomatosis	Liver/biliary system, homeostasis/metabolism	Not known

<i>DAB2</i>	1	No OMIM	renal/urinary system, lethality-prenatal/perinatal, homeostasis/metabolism, growth/size, embryogenesis	Not known
<i>DICER1</i>	1	No OMIM	liver/biliary system, limbs/digits/tail, life span-post-weaning/aging, lethality-postnatal, lethality-prenatal/perinatal, homeostasis/metabolism, growth/size, embryogenesis, cellular, cardiovascular system, behavior/neurological, immune system, respiratory system, skeleton, touch/vibrissae, skin/coat/nails, hematopoietic system	Not known
<i>DLL3</i>	1	Spondylocostal dysostosis, autosomal recessive	nervous system, muscle, limbs/digits/tail, lethality-postnatal, hearing/vestibular/ear, growth/size, embryogenesis, craniofacial, behavior/neurological, reproductive system, skeleton, vision/eye	Not known
<i>DOCK1</i>	1	No OMIM	muscle, lethality-postnatal, lethality-prenatal/perinatal, homeostasis/metabolism, growth/size, embryogenesis	Not known
<i>DSCAM</i>	6	No OMIM	nervous system, life span-post-weaning/aging, behavior/neurological, skeleton, vision/eye, respiratory	respiratory, nervous system, reproductive system
<i>EP400</i>	4	No OMIM	nervous system, lethality-prenatal/perinatal, embryogenesis, hematopoietic system	Not known
<i>ERC2</i>	1	No OMIM	nervous system	Not known
<i>ESAM</i>	1	No OMIM	growth/size, cardiovascular system, immune system	Not known
<i>ESX1</i>	1	No OMIM	growth/size, embryogenesis	growth/size, embryogenesis
<i>F7</i>	1	Factor VII deficiency	Normal, lethality-prenatal/perinatal, homeostasis/metabolism, cardiovascular system	Normal
<i>FAM48A</i>	1	No OMIM	pigmentation, nervous system, lethality-prenatal/perinatal, growth/size, embryogenesis, cellular, vision/eye	Not known
<i>FN1</i>	2	Glomerulopathy with fibronectin deposits 2	tumorigenesis, normal, nervous system, muscle, life span-post-weaning/aging, lethality-prenatal/perinatal, homeostasis/metabolism, growth/size, embryogenesis, cellular,	Not known

			cardiovascular system, immune system, reproductive system, skeleton, other, hematopoietic system	
<i>GBA</i>	2	Gaucher disease; Parkinson's disease	Nervous system, liver/biliary system, life span-post-weaning/aging, lethality-postnatal, lethality-prenatal/perinatal, homeostasis/metabolism, growth/size, cardiovascular system, behavior/neurological, immune system, respiratory system, skeleton, skin/coat/nails, hematopoietic system	Homeostasis/metabolism
<i>GLT8D1</i>	1	No OMIM	normal	
<i>GPHB5</i>	1	No OMIM	normal	
<i>GRIN3B</i>	1	No OMIM	growth/size, behavior/neurological	Not known
<i>GRIP2</i>	2	No OMIM	normal	
<i>HADHB</i>	2	Trifunctional protein deficiency	muscle, liver/biliary system, life span-post-weaning/aging, adipose tissue, homeostasis/metabolism, growth/size, cardiovascular system	Not known
<i>INSR</i>	1	Leprechaunism; Rabson-Mendenhall syndrome; Diabetes mellitus, insulin-resistant, with acanthosis nigricans; Hyperinsulinemic hypoglycemia, familial, 5	Nervous system, renal/urinary system, muscle, liver/biliary system, life span-post-weaning/aging, lethality-postnatal, adipose tissue, Homeostasis/metabolism, growth/size, endocrine/exocrine gland, digestive/alimentary, cellular, cardiovascular system, behavior/neurological, respiratory system, reproductive system, hematopoietic system	Homeostasis/metabolism, digestive/alimentary, endocrine/exocrine
<i>IRAK3</i>	1	Asthma susceptibility	Limbs/digits/tail, growth/size, immune system, skeleton, hematopoietic system	Not known
<i>KLF4</i>	1	No OMIM	tumorigenesis, lethality-prenatal/perinatal, homeostasis/metabolism, digestive/alimentary, behavior/neurological, skin/coat/nails	Not known
<i>KRT14</i>	7	Epidermolysis bullosa simplex; Naegeli-Franceschetti-Jadassohn syndrome; Dermatopathia pigmentosa reticularis,	Pigmentation, tumorigenesis, life span-post-weaning/aging, lethality-postnatal, digestive/alimentary, respiratory system, reproductive system, vision/eye, skin/coat/nails	Not known

<i>LHB</i>	1	Hypogonadism, hypergonadotropic; Male pseudohermaphroditism	homeostasis/metabolism, endocrine/exocrine gland, reproductive system	Not known
<i>LRP2</i>	1	Donnai-Barrow syndrome	Nervous system, renal/urinary system, lethality-prenatal/perinatal, homeostasis/metabolism, hearing/vestibular/ear, growth/size, endocrine/exocrine gland, embryogenesis, craniofacial, cellular, respiratory system, reproductive system, skeleton, vision/eye	Hearing/vestibular/ear
<i>MAP3K14</i>	1	No OMIM	immune system	Not known
<i>MAP7</i>	1	No OMIM	endocrine/exocrine gland, reproductive system	Not known
<i>MAPK7</i>	1	No OMIM	nervous system, muscle, life span-post-weaning/aging, lethality-prenatal/perinatal, homeostasis/metabolism, hearing/vestibular/ear, growth/size, embryogenesis, digestive/alimentary, craniofacial, cellular, cardiovascular system, behavior/neurological, skin/coat/nails	Not known
<i>MLL3</i>	1	No OMIM	growth/size, cellular, reproductive system, vision/eye	Not known
<i>MMP12</i>	2	No OMIM	nervous system, homeostasis/metabolism, immune system, reproductive system	Not known
<i>MYO6</i>	1	Deafness, autosomal dominant 22; Deafness, autosomal recessive 37; Deafness, sensorineural, with hypertrophic cardiomyopathy	Nervous system, hearing/vestibular/ear, growth/size, behavior/neurological, reproductive system, vision/eye	Hearing/vestibular/ear
<i>MYT1</i>	1	No OMIM	muscle, lethality-prenatal/perinatal, homeostasis/metabolism, endocrine/exocrine gland, digestive/alimentary	Not known
<i>NBN</i>	2	Nijmegen breakage syndrome; Leukemia, acute lymphoblastic	tumorigenesis, nervous system, life span-post-weaning/aging, lethality-prenatal/perinatal, growth/size, endocrine/exocrine gland, embryogenesis, cellular, behavior/neurological, immune system, reproductive system, hematopoietic system	Cellular, life span-post-weaning/aging, tumorigenesis. Human disease model.

<i>NCOA3</i>	2	No OMIM	lethality-postnatal, homeostasis/metabolism, hearing/vestibular/ear, growth/size, endocrine/exocrine gland, embryogenesis, craniofacial, cellular, reproductive system, skin/coat/nails	Not known
<i>NEFM</i>	1	No OMIM	nervous system	Not known
<i>NR2E3</i>	1	Enhanced S-cone syndrome; Retinitis pigmentosa-37	Nervous system, vision/eye	Not known
<i>NTSR1</i>	1	No OMIM	nervous system, adipose tissue, homeostasis/metabolism, growth/size, digestive/alimentary, behavior/neurological, touch/vibrissae	digestive/alimentary
<i>PADI6</i>	1	No OMIM	embryogenesis, reproductive system	Not known
<i>PATZ1</i>	1	No OMIM	endocrine/exocrine gland, reproductive system	Not known
<i>PCDH8</i>	1	No OMIM	normal	
<i>PERP</i>	1	No OMIM	lethality-postnatal, cellular	Not known
<i>PFN2</i>	1	No OMIM	nervous system, cellular, behavior/neurological	Not known
<i>PIM2</i>	1	No OMIM	immune system, hematopoietic system	Not known
<i>PKD1L3</i>	1	No OMIM	Normal	
<i>PNLIPRP2</i>	2	No OMIM	homeostasis/metabolism, growth/size, digestive/alimentary, immune system	Not known
<i>PNOC</i>	1	No OMIM	homeostasis/metabolism, behavior/neurological, touch/vibrissae	Not known
<i>PNPLA2</i>	1	Neutral lipid storage disease with myopathy	muscle, liver/biliary system, life span-post-weaning/aging, adipose tissue, homeostasis/metabolism, growth/size, cardiovascular system	Not known
<i>POU2F1</i>	1	No OMIM	lethality-prenatal/perinatal, vision/eye, hematopoietic system	Not known
<i>PPP1R9B</i>	2	No OMIM	nervous system, homeostasis/metabolism, growth/size, behavior/neurological	Not known
<i>PRKCH</i>	1	Cerebral infarction susceptibility	Tumorigenesis, homeostasis/metabolism, skin/coat/nails	Not known
<i>PRKDC</i>	2	No OMIM	tumorigenesis, life span-post-weaning/aging, homeostasis/metabolism, endocrine/exocrine gland, digestive/alimentary, cellular, immune	Not known

			system, hematopoietic system	
<i>PTK7</i>	1	No OMIM	nervous system, renal/urinary system, limbs/digits/tail, lethality-prenatal/perinatal, hearing/vestibular/ear, embryogenesis	Not known
<i>RAB3IP</i>	1	No OMIM	normal	
<i>REC8</i>	1	No OMIM	lethality-prenatal/perinatal, growth/size, endocrine/exocrine gland, reproductive system	Not known
<i>RECQL4</i>	1	Rothmund-Thomson syndrome; RAPADILINO syndrome; Baller-Gerold syndrome	Pigmentation, tumorigenesis, limbs/digits/tail, life span-post-weaning/aging, lethality-postnatal, lethality-prenatal/perinatal, growth/size, embryogenesis, digestive/alimentary, craniofacial, cellular, immune system, skeleton, vision/eye, skin/coat/nails, hematopoietic system	Not known
<i>RETNLB</i>	2	No OMIM	life span-post-weaning/aging, homeostasis/metabolism, digestive/alimentary, immune system	Not known
<i>RHBG</i>	1	No OMIM	normal	
<i>RIPK2</i>	1	No OMIM	immune system, hematopoietic system	Not known
<i>RP1</i>	1	Retinitis pigmentosa-1	Nervous system, vision/eye	Vision/eye
<i>RYR3</i>	1	No OMIM	nervous system, muscle, behavior/neurological	behavior/neurological
<i>SARM1</i>	4	No OMIM	cellular	Not known
<i>SEBOX</i>	1	No OMIM	Normal	
<i>SETD8</i>	1	No OMIM	lethality-prenatal/perinatal	Not known
<i>SFRS1</i>	1	No OMIM	lethality-prenatal/perinatal	Not known
<i>SLC18A1</i>	1	No OMIM	normal	
<i>SOX6</i>	1	No OMIM	normal, limbs/digits/tail, lethality-postnatal, lethality-prenatal/perinatal, growth/size, respiratory system, skeleton	Not known
<i>ST3GAL1</i>	1	No OMIM	Immune system	Not known
<i>STK17B</i>	1	No OMIM	immune system, hematopoietic system	Not known
<i>TAP2</i>	1	Bare lymphocyte syndrome, type I; Wegener-like granulomatosis	Immune system, hematopoietic system	Not known
<i>TCOF1</i>	1	Treacher Collins mandibulofacial dysostosis	Nervous system, lethality-prenatal/perinatal, hearing/vestibular/ear, growth/size, endocrine/exocrine gland, embryogenesis,	Nervous system, lethality-prenatal/perinatal, hearing/vestibular/ear,

			digestive/alimentary, craniofacial, cellular, respiratory system, skeleton, vision/eye, taste/olfaction	growth/size, endocrine/exocrine gland, embryogenesis, digestive/alimentary, craniofacial, cellular, respiratory system, skeleton, vision/eye, taste/olfaction. Human disease model.
<i>TEKT4</i>	6	No OMIM	reproductive system	Not known
<i>THBS2</i>	1	Lumbar disc herniation susceptibility	Tumorigenesis, muscle, limbs/digits/tail, life span-post-weaning/aging, lethality-prenatal/perinatal, homeostasis/metabolism, cellular, cardiovascular system, behavior/neurological, immune system, respiratory system, skeleton, skin/coat/nails, hematopoietic system	Not known
<i>THBS4</i>	1	No OMIM	normal	
<i>TYRO3</i>	1	No OMIM	nervous system, behavior/neurological	Not known
<i>ZFX</i>	2	No OMIM	lethality-postnatal, lethality-prenatal/perinatal, growth/size, endocrine/exocrine gland, embryogenesis, behavior/neurological, reproductive system	lethality-postnatal, lethality-prenatal/perinatal, reproductive system
<i>ZPBP</i>	1	No OMIM	endocrine/exocrine gland, reproductive system	reproductive system