Gene and/or mutant	Protein Function	NTD Type(s)	Penetrance	References
Efna5	Cell Adhesion & Interaction	EX or SF+EX	20% (F:M=2:1)	Holmberg et al. (2000), Abdul-Aziz et al. (2009)**
Epha7	Cell Adhesion & Interaction	EX	25%	Holmberg et al. (2000), Abdul-Aziz et al. (2009)**
Epha2tm1Jrui/+Epha4rb-2J/+ double heterozygous	Cell Adhesion & Interaction	SBO or Ex	28% or 16%	Abdullah et al. (2017)
Frem1	Cell Adhesion & Interaction	EX	2%	Varnum & Fox (1981); Smyth et al. (2004)
Frem2 (my mutant; my-F11 ENU mutant)	Cell Adhesion & Interaction	EX	5%- most	Carter (1959); Jadeja et al. (2005); Timmer et al. (2005)
Gja1, Gja5 (Cx43, Cx40; digenic)	Cell Adhesion & Interaction	EX	17% -/-,-/-; 5% +/-,-/-; 2% +/+,-/-	Lo et al. (1997); Simon et al. (2004)
Grip1(eb mutant)	Cell Adhesion & Interaction	EX	30%	Swiergiel et al. (2000); Takamiya et al. (2004)
Hspg2	Cell Adhesion & Interaction	EX	6%	Arikawa-Hirasawa et al. (1999)
Itga3, Itga6 (digenic)	Cell Adhesion & Interaction	EX	45%	De Arcangelis et al. (1999); Thorsteinsdottir et al. (1995)
Itgb1 (a isoform)	Cell Adhesion & Interaction	EX or SBA	some	Baudoin et al. (1998); Ohyama et al. (1997)
Conditional Itgb1-/- in surface ectoderm	Cell Adhesion & Interaction	SBA	56%	Mole et al. (2020)
Conditional Itgb1-/- in neural ectoderm	Cell Adhesion & Interaction	EX or SBA	9% or 9%	Mole et al. (2020)
Lama5 (ENU)	Cell Adhesion & Interaction	EX	no data	Zohn et al. (2005)
Lama5 null	Cell Adhesion & Interaction	EX	60%	Miner et al. (1998)
*Lmnb1***	Cell Adhesion & Interaction	Modifier of ct (Grhl3)	N/A	De Castro et al. (2012)
Piga	Cell Adhesion & Interaction	EX + SF	75% in heterozygotes (-/- die by GD10)	Nozaki et al. (1999)
Rgma	Cell Adhesion & Interaction	EX	50%	Niederkofler et al. (2004)
Myo10 null	Cell Adhesion & Interaction	Ex or CRS	73% or 18%	Heimsath et al.(2017)
Gadd45a	Cell cycle	EX	10%	Hollander et al. (1999)
Nap1l2 chimera	Cell cycle	EX	some	Rogner et al. (2000)
*Nls1***	Cell cycle	EX		Regeling et al. (2011)
Phactr4 (humdy mutant) (defect in PP1 binding)	Cell cycle	EX	100%	Kim et al. (2007)
*Rad9b ** *	Cell cycle	EX	20% in heterozygotes (-/- die by GD10)	Leloup et al. (2010)
Terc	Cell cycle	SF, EX, SBA	10% SF, 15% EX, 5% SBA	Herrera et al. (1999)
Tsc1	Cell cycle	EX	15%	Wilson et al. (2005)
Tsc2	Cell cycle	EX	50%	Kobayashi et al. (1999)
Xrcc2	Cell cycle	EX	30%	Deans et al. (2000)
Enpp2 (ATX)	Cell migration	open neural tube	100%	Fotopoulu et al. (2010), van Meeteren et al. (2006)
*Plxnb2***	Cell migration	EX +/- SBA	90% EX, 45% SBA or CT	Deng et al. (2007); Friedel et al. (2007)
*Sema4c***	Cell migration	EX	35%	Maier et al. (2010)
Ambra1	Cell survival	EX +/or SBA	on B6: 15% EX, 65% SBA, 20% EX+SBA (on mixed: 50% EX, 5% SBA, 30% EX+SBA)	Fimia et al. (2007)
Apaf1 (fog mutant)	Cell survival	SBA +/or SF+rostral EX	40% SBA; 10% SF+EX	Harris et al. (1997); Honarpour et al. (2001)
Apaf1 null	Cell survival	SF+rostral EX; hindbrain EX	35% SF+EX; 35% hindbrain EX	Cecconi et al. (1998); Yoshida et al. (1998)
Bcl10	Cell survival	hindbrain EX	30%	Ruland et al. (2001)
Casp3	Cell survival	SF+rostral EX or hindbrain EX	100%	Houde et al. (2004); Urase et al. (2003)
Casp9	Cell survival	hindbrain EX	some	Hakem et al. (1998)
Chuk, Ikbkb (digenic)	Cell survival	hindbrain EX	70%	Li et al. (2000)
Cycs (Cyt c; null for apoptotic function)	Cell survival	EX	25%	Hao et al. (2005)
Enpp2 (ATX)	Cell survival	open neural tube	100%	Fotopoulu et al. (2010), van Meeteren et al. (2006)
Map3k4 null	Cell survival	EX +/or SBA	20% EX+SBA, 35% EX, 35% SBA	Chi et al. (2005)
Map3k4 partial function	Cell survival	EX +/or SBA	50% EX +/or SBA	Abell et al. (2005)
Mapk8, Mapk9 (Jnk1, Jnk2; digenic)	Cell survival	hindbrain EX	100% -/-, -/- (25% -/-,+/-)	Kuan et al. (1999); Sabapathy et al. (1999)
Mdm4	Cell survival	“improper neural tube closure”	all?	Migliorini et al. (2002)
*Nls1***	Cell survival	EX	¾ females, 1/6 males	Regeling et al. (2011)
Por (CYPOR)	Cell survival	EX	some EX	Shen et al. (2002)
Ptdsr	Cell survival	EX	2-15%	Li et al. (2003); Bose et al. (2004)
*Rad9b ** *	Cell survival	EX	20% in heterozygotes (-/- die by GD10)	Leloup et al. (2010)
Sphk1, Sphk2 (digenic)	Cell survival	EX	20%	Mizugishi et al. (2005)
Terc	Cell survival	SF, EX, SBA	10% SF, 15% EX, 5% SBA	Herrera et al. (1999)
Traf4	Cell survival	SBA	some	Masson et al. (1998); Regnier et al. (2002)
Traf6	Cell survival	EX	35%	Lomaga et al. (2000)
Txn2	Cell survival	EX	all	Nonn et al. (2003)
Xrcc2	Cell survival	EX	30%	Deans et al. (2000)

Apob null	Cellular transport	EX	all alive past GD10	Farese et al. (1995)
Apob truncated	Cellular transport	EX	30%	Homanics et al. (1995)
Atp2c1 (Spca1)	Cellular transport	EX	all	Okunade et al. (2007)
Atp2c1 (Spca1) 1D mutant	Cellular transport	EX or SBO or CRS	100% or 100% or 2.4%	Brown et al. (2018)
Fpn1 (Slc40a1) (hypomorph)	Cellular transport	EX +/- SBA	100% EX, 1% SBA on C3HxC57BL/6; 0% on 129	Zohn et al. (2007)
Mttp	Cellular transport	EX	all	Raabe et al. (1998)
Ptpn9 (Meg2)	Cellular transport	EX +/- SBA	98% EX; 50% SBA	Wang et al. (2005)
Rab23 (opb mutant)	Cellular transport	EX +/- SBA	2-100% EX; 1-2 % SBA	Gunther et al. (1994)
Rab23 (opb2 ENU mutant)	Cellular transport	EX +/- SBA	no data	Kasarskis et al. (1998); Eggenschwiler et al. (2001)
Sec24b	Cellular transport	CRS	100%	Merte et al. (2010)
Snx1, Snx2 (digenic)	Cellular transport	EX	20% in +/-; -/-	Griffin et al. (2005)
Snx13	Cellular transport	EX	100%	Zheng et al. (2006)
Trpm6	Cellular transport	EX	~30% (some SBO)	Walder et al. (2009)
Cobl hypomorph; Lp/+ (digenic)	Cilia	EX	20%	Carroll et al. (2003)
Dvl2	Cilia	thoracic SBA+EX	3%	Klingensmith et al. (1996); Hamblet et al. (2002)
*Arl13b (hennin ENU mutant)***	Cilia	EX + SBA	all?	Horner et al. (2011); Caspary et al. (2007)
*B9d1***	Cilia	EX	6%	Dowdle et al. (2011)
Bbs4	Cilia	EX	15%	Ross et al. (2005)
*Tctn2***	Cilia	EX	100% (on 129xC57BL/6)	Sang et al. (2011)
Dync2h1 (lln mutant)	Cilia	EX	50%	Huangfu & Anderson (2005)
Dync2li1	Cilia	EX	all	Rana et al. (2004)
Ift172 (wim mutant)	Cilia	EX	all?	Huangfu et al. (2003)
*Traf3ip1***	Cilia	EX	some (% not given)	Berbari et al. (2011)
*bromi (ENU#10)***	Cilia	EX	some?	Ko et al. (2010); Zohn et al. (2005)
C2cd3 (Hty mutant)	Cilia	EX	30%	Hoover et al. (2008)
*Cluap1***	Cilia	“unclosed NT”	60%?	Botilde et al. (2013); Pasek et al. (2012)
Ift122	Cilia	EX	all?	Cortellino et al. (2009)
*Ift140 (cauli mutant)***	Cilia	EX (+SF) +/- SBA	100% EX(+SF?),some SBA	Miller et al. (2013)
Kif3b	Cilia	EX	all?	Nonaka et al. (1998)
Inpp5e	Cilia	EX	30%	Jacoby et al. (2009)
Fuz	Cilia	EX	“few”	Gray et al. (2009); Heydeck et al. (2009); Heydeck and Liu (2011)
Intu	Cilia	EX	30%	Zeng et al. (2010)
Mks1 (krc mutant)	Cilia	EX	30%	Weatherbee et al. (2009)
Stil-/-	Cilia	Cranial NTD	all?	Izraeli et al. (1999)
*Tmem67***	Cilia	EX, frontal encephalocele	25% EX, 25% encephalocele	Abdelhamed et al. (2013)
*Tmem107***	Cilia	EX	xxx	Christopher et al. (2012)
vl mutant	Cilia	SBA	40%	Wilson & Wyatt (1992)
Sec24b	Polarity	CRS	100%	Merte et al. (2010)
*Cdx1, Cdx2** (digenic)*	Polarity	CRS	100%	Savory et al. (2011)
Celsr1 (Scy & Crsh mutants)	Polarity	CRS	100%	Curtin et al. (2003)
Cthrc1-/-;Vangl2-Lp/+ (digenic)	Polarity	EX	all	Durmus et al. (2006); Yamamoto et al. (2008)
Dvl1, Dvl2 (digenic)	Polarity	CRS	95%	Hamblet et al. (2002)
Dvl2+/-,Dvl3-/- (digenic)	Polarity	CRS	some	Etheridge et al. (2008)
Dvl2-/-,Dvl3+/- (digenic)	Polarity	CRS	~100%	Etheridge et al. (2008)
*Fat1, Fat4***	Polarity	EX	75%	Saburi et al. (2012)
*Fat1, Fjx1***	Polarity	EX	50%	Saburi et al. (2012)
Fat1-/-	PCP	Ex	40%	Badouel et al. (2015)
Fzd3, Fzd6 (digenic)	Polarity	CRS	most	Wang et al. (2006)
Ptk7	Polarity	CRS, occasional SBA	all	Lu et al. (2004); Paudyal et al. (2010)**
Scrib (Crc mutant)	Polarity	CRS	100%	Murdoch et al. (2001, 2003)
*Sdc4-/-; Lp/+***	Polarity	SBA	55%	Escobedo et al. (2013)
*Sestd1***	Polarity	caudal regression syndrome +/- SBA (like Dact1)	“partially penetrant”	Yang et al. (2013)
Sfrp1,Sfrp2,Sfrp5 (trigenic)	Polarity	CRS	100%	Satoh et al. (2008)
Smurf1,Smurf2	Polarity	CRS	65%	Narimatsu et al. (2009)
Vangl2 (Lp mutant)	Polarity	CRS, occasional SBA in Lp/+	100%	Murdoch et al. (2001a); Kibar et al. (2001)

Lp, Crc mutants (Vangl2, Scrib) digenic, heterozygous	Polarity	CRS or SBA	55% CRS, 5% SBA	Murdoch et al. (2001b)
Lp, Crsh mutants (Vangl2, Celsr1) digenic, heterozygous	Polarity	CRS	no data	Copp et al. (2003b)
Lp, Ptk7 (digenic, heterozygous)	Polarity	SBA	95%	Lu et al. (2004)
Vangl1+/-, Vangl2-Lp/+ (digenic)	Polarity	CRS	60%	Torban et al. (2008)
Vangl2-Lp/+, Dvl2-/- (digenic)	Polarity	CRS	100%	Wang et al. (2006)
Vangl2-Lp/+, Dvl3+/- (digenic)	Polarity	CRS, EX	some	Etheridge et al. (2008)
Vangl2-Lp/+, Sec24b+/- (digenic)	Polarity	SBA	70%	Merte et al. (2010)
Vangl2-Lp/+, Sfrp1-/-, Sfrp2+/-, Sfrp5-/- (4 genes)	Polarity	SBA	60%	Satoh et al. (2008)
*Dlgh1***	Polarity	CRS	7% (plus open eyelids, CP, cochlear defects)	Rivera et al. (2013)
*Wnt5a***	Polarity	CRS or EX	5%	Yamaguchi et al. (1999); Qian et al. (2007); Schleiffarth et al. (2007); He et al. (2008)
*Dact1***	Polarity	caudal regression syndrome (not NTD) +/- SBA	95% caudal regression, 15-20% SBA	Suriben et al. (2009), Wen et al. (2010a)
Abi1	Cytoskeleton	no closure	all	Rakeman & Anderson (2006)
Abl1, Abl2 (digenic)	Cytoskeleton	EX	some	Koleske et al. (1998)
Cfl1	Cytoskeleton	no closure	all	Gurniak et al. (2005)
Dlc1	Cytoskeleton	EX	all?	Durkin et al. (2005)
Enah, Pfn1(-/-, +/-) (digenic)	Cytoskeleton	EX	50%	Lanier et al. (1999)
Enah, Vasp (digenic)	Cytoskeleton	EX, some SF+EX	-/-,-/- 64% EX some SF; +/-,-/- 18% EX; -/-,+/- 7% EX	Menzies et al. (2004)
Grlf1 (p190RhoGAP)	Cytoskeleton	EX	30%	Brouns et al. (2000)
Marcks	Cytoskeleton	EX	30% in females; 15% in males	Stumpo et al. (1995); Blackshear et al. (1996)
Marcksl1 (Mlp)	Cytoskeleton	EX +/or SBA	55-100% EX; 0-15% SBA	Chen et al. (1996); Wu et al. (1996)
Palld	Cytoskeleton	EX	100%	Luo et al. (2005)
Pip5k1c	Cytoskeleton	EX	100%	Wang et al. (2007)
Rac1-/-, Grhl3-Cre/+ (digenic) (Rac1 is conditional null in expression domain of Grhl3)	Cytoskeleton	EX +/or SBA	60% EX, 85% SBA	Camerer et al. (2010); Sugihara et al. (1998)
Rac1-/-, Grhl3-Cre/+ (digenic) (Rac1 is conditional null in expression domain of Grhl3)	Cytoskeleton	EX +/or SBA	60% EX, 85% SBA	Camerer et al. (2010); Sugihara et al. (1998)
Conditional Rac1f/f or f/- with Pax3Cre/+	Cytoskeleton	SBA(curlied tail) or EX	76% or 8%	Rolo et al. (2016)
Conditional Rac1f/f or f/- with Grhl3Cre/+	Cytoskeleton	SBA(curlied tail) or EX	89% or 25%	Rolo et al. (2016)
Shroom3	Cytoskeleton	EX +/- SF, +/or SBA	100% EX; 85% SF; 25% SBA	Hildebrand & Soriano (1999)
Snx13	Cytoskeleton	EX	100%	Zheng et al. (2006)
Vcl	Cytoskeleton	SF + EX	all	Xu et al. (1998)
*Alkbh1***	Epigenetic Regulator	EX	2% (10% of the 20% alive on GD10-11)	Ougland et al. (2012)
Cecr2	Epigenetic Regulator	EX	0-75%	Banting et al. (2005)
Dnmt3b	Epigenetic Regulator	EX	most or all	Okano et al. (1999)
Dnmt3l (+/- embryos in -/- dam)	Epigenetic Regulator	EX	15%	Hata et al. (2002)
*Fbxl10***	Epigenetic Regulator	EX	50%	Fukuda et al. (2011)
Hdac4	Epigenetic Regulator	EX	some	Vega et al. (2004)
*Jarid1b***	Epigenetic Regulator	EX	5%	Albert et al. (2013)
Kat2a (Gcn5) (conditional KO, hypomorph)	Epigenetic Regulator	EX	15% in fn/fn; 100% in fn/null on 129, 20% on C57BL/6	Bu et al. (2007); Lin et al. (2008); Xu et al. (2000)
*Ppm1g***	Epigenetic Regulator	EX	30%	Foster et al. (2013)
Sirt1	Epigenetic Regulator	EX	some	Cheng et al. (2003)
Smarca4 heterozygotes	Epigenetic Regulator	EX	15% in heterozygotes	Bultman et al. (2000); Randazzo et al. (1994)
*Smarcc1 (Baf155)** hypomorph*	Epigenetic Regulator	EX	80% in homozygotes (heterozygotes normal)	Harmacek et al. (2013 or 2014)
Suz12+/-	Epigenetic Regulator	SB	incomplete penetrance	Miro et al. (2009)
Smarcc1 heterozygotes	Epigenetic Regulator	EX	20% in heterozygotes	Kim et al. (2001)
*Utx, Uty** (homologous)*	Epigenetic Regulator	EX	all	Westead et al. (2012), Shpargel et al. (2012)
*Amt***	Metabolism	EX or CRS	82% EX, 5% CRS	Narisawa et al. (2011)
GldcGT2/GT2	Metabolism	Primarily EX, some SBA or CRS	53%	Leung et al. (2017)
Folr1 null	Metabolism	no closure	all	Piedrahita et al. (1999)
Folr1 partial rescue by folate	Metabolism	EX	5-30%	Piedrahita et al. (1999); Saitsu et al. (2003); Spiegelstein et al. (2004a, 2004b)
*Mthfd1l***	Metabolism	EX or CRS	55% EX, 5% CRS (1/26)	Momb et al. (2013)

*Mtrr hypomorph***	Metabolism	EX	5% in grandprogeny of a maternal grandfather or grandmother gt/+; independent of embryo genotyoe	Padmanabhan et al. (2013)
Nat2	Metabolism	EX	2%	Wakefield et al. (2007, 2008)
Pip5k1c	Metabolism	EX	100%	Wang et al. (2007)
Por (CYPOR)	Metabolism	EX	some EX	Shen et al. (2002)
Conditional Slc19a-/- in whole embryo, or neural crest cell, or yolk sac	Metabolism	EX	71.4% or 34.6% or 33.3%	Toriyama et al. (2017)
Slc25a19	Metabolism	EX	all	Lindhurst et al. (2006)
Slc25a32gt/gt	Metabolism	Majority Ex and some CRS	100%	Kim et al. (2018)
Aldh1a2	Signaling	Craniorachischisis	all	Niederreither et al. (1999)
*Alx3***	Signaling	EX (+/-SF)	60% EX, some SF	Lakhwani et al. (2010)
Apob null	Signaling	EX	all alive past GD10	Farese et al. (1995)
Apob truncated	Signaling	EX	30%	Homanics et al. (1995)
Arnt	Signaling	SF, EX, SF+EX	40%	Kozak et al. (1997)
Axin1(Fu mutant)	Signaling	EX or SBA	10%EX, 25%SBA	Theiler & Gluecksohn-Waelsch (1956); Zeng et al. (1997)
Birc5 (survivin) conditional null in endothelial cells	Signaling	EX	30%	Zwerts et al. (2007)
Bmp2 conditional null, flox(neo), hypomorph	Signaling	EX	25% in flox/null	Singh et al. (2008)
Bmp2 null	Signaling	EX	60% in heterozygote (-/- die by GD10)	Castriano & Mishina (2009)
Bmp5, Bmp7 (digenic)	Signaling	EX	all	Solloway & Robertson (1999)
Brca1 hypomorph	Signaling	EX	10%	Xu et al. (2001)
Brca1 null	Signaling	EX +/or SBA	40%	Gowen et al. (1996)
Calr	Signaling	EX	15%	Rauch et al. (2000)
Cart1	Signaling	EX	65-100%	Zhao et al. (1996)
Cited2	Signaling	EX	60-80%	Dunwoodie et al. (1998); Bamforth et al. (2001); Barbera et al. (2002)
Crebbp	Signaling	EX	all	Tao et al. (1998)
Csk	Signaling	EX	all	Imamoto et al. (1993); Nada et al. (1993)
Csnk2a1	Signaling	EX	>90%	Lou et al. (2008); Seldin et al. (2008)
*Ctnnb1 (b catenin conditional KO)***	Signaling	SBA	100%	Zhao et al. (2014)
Ctnnbip1 (ICAT)	Signaling	EX+SF	15%	Satoh et al. (2004)
Cyp26a1	Signaling	SBA +/- EX	most SBA, some EX	Abu-Abed et al. (2001, 2003)
Deaf1	Signaling	EX	80%	Hahm et al. (2004)
Dlx5	Signaling	EX	30%	Depew et al. (1999)
Edg2	Signaling	EX	5%	Contos et al. (2000)
Ep300	Signaling	EX	100%	Yao et al. (1998)
Epor	Signaling	hindbrain EX	all?	Tsai et al. (2006)
F2r, F2rl1 (Par1, Par2) (digenic)	Signaling	EX +/or SBA	35% EX, rare SBA	Camerer et al. (2010), Copp & Greene (2010)
Fdft1	Signaling	EX	60%	Tozawa et al. (1999)
Fgfr1 (a isoform)	Signaling	SBA	all	Xu et al. (1999)
Fgfr1 chimera	Signaling	SBA	70%	Deng et al. (1997); Yamaguchi et al. (1992)
Fgfr2C342Y	Signaling	Ex or SBA	63% or <1%	Peskett et al. (2017)
Fkbp1a	Signaling	EX	10%	Shou et al. (1998)
Fkbp8 hypomorph	Signaling	SBA	100%	Finnell et al. (2006); Bulgakov et al. (2004)
*Fkbp8 null***	Signaling	SBA +/- EX	100% SBA; some EX	Shirane et al. (2008); Shirane-Kitsuji and Nakayama (2014)
Foxb1	Signaling	EX	5%	Labosky et al. (1997)
Fzd3	Signaling	EX	25%	Borello et al. (1999); Wang et al. (2002)
Gli3 (Xt, Bph, Pdn mutants)	Signaling	EX	20-60%	Johnson (1969); Hui & Joyner (1993); Sasaki et al. (1997)
Gnaz-/-, Grhl3-Cre/+ (digenic) (Gnaz is conditional null in domain of Grhl3; the 4 other Gi/o/z genes inactivated in Grhl3 domain by pertussis toxin)	Signaling	EX +/or SBA	20% EX, 40% SBA	Camerer et al. (2010); Hendry et al. (2000)
Gnb1	Signaling	EX	40%	Okae & Iwakura (2010)
Gpr161 (vl mutant, hypomorph)	Signaling	SBA	40%	Korstanje et al. (2008); Matteson et al. (2008)
*Grhl2 (Axd mutant, hypermorph)***	Signaling	SBA	40-100% (F:M=1:1)	Brouns et al. (2011); Essien et al. (1990)
*Grhl2 (null)***	Signaling	EX+SF, SBA	100% EX+SF, 90% SBA (15% EX+SF in heterozygote)	Brouns et al. (2011); Rifat et al. (2010); Werth et al. (2010)
*Grhl21Nis (ENU mutant, truncated)***	Signaling	EX+SF	100%	Pyrgaki et al. (2011)
Grhl3 null	Signaling	SBA +/- EX	100% SBA, 2% EX	Ting et al. (2003); Auden et al. (2006)

Gtf2i	Signaling	EX	60% in null, 15% in +/-	Enkhmandakh et al. (2009)
Gtf2ird1	Signaling	EX	10% in null, 2% in +/-	Enkhmandakh et al. (2009)
Hectd1 (opm mutant)	Signaling	EX	90% in opm/opm, 5% in opm/+	Zohn et al. (2007); Kasarskis et al. (1998); Sarkar and Zohn (2012)
Hes1	Signaling	SF+EX	70%	Ishibashi et al. (1995); Lee et al. (2005a)
Hic1	Signaling	EX	35%	Carter et al. (2000)
Hif1a	Signaling	EX	all	Iyer et al. (1998)
Hipk1, Hipk2 (digenic)	Signaling	EX (F:M=2:1)	50% -/-,-/-; 10%+/-,-/-; 5%-/-,+/-	Isono et al. (2006); Aikawa et al. (2006)
Hmx1 (dmbo mutant)	Signaling	EX	25%	Munroe et al. (2009)
Hspg2	Signaling	EX	6%	Arikawa-Hirasawa et al. (1999)
Inka1	Signaling	EX	5%	Reid et al. (2010)
Itpk1(hypomorph)	Signaling	EX +/or SBA	10%	Wilson et al. (2009)
Jarid2 (jmj mutant)	Signaling	EX	50%	Takeuchi et al. (1995)
Klkb1	Signaling	No closure	all	Ylikorkala et al. (2001)
Lmo4	Signaling	EX	10-50%	Hahm et al. (2004); Tse et al. (2004); Lee et al. (2005b)
*Lrp2***	Signaling	EX	40%	Kur et al. (2014)
Lrp6	Signaling	EX +/or SBA	50% EX +/or SBA	Pinson et al. (2000)
Lrp6 (Cd mutant, hypermorph)	Signaling	EX	20% (F:M=2:1)	Carter et al. (2005)
Lrp6 (rs mutant, hypomorph)	Signaling	SBA	some	Kokubu et al. (2004)
*Lrp6Skax26-Jus hypermorph***	Signaling	Curly tail	10-50% in -/-; 5-10% in +/-	Allache et al. (2013)
Luzp1	Signaling	EX	40%	Hsu et al. (2008)
Map3k7	Signaling	hindbrain EX	most	Shim et al. (2005)
Med12	Signaling	EX or SBA or CRS	female mosaic hemizygotes: 5% EX, 65% EX+SBA, 30% CRS	Rocha et al. (2010)
Men1	Signaling	EX	20% on 129, 0% on C57BL/6	Bertolino et al. (2003); Lemos et al. (2009)
Mib2	Signaling	EX	10-50%	Wu et al. (2007)
Msgn1	Signaling	SBA	all?	Yoon & Wold (2000)
Msx1, Msx2 (digenic)	Signaling	EX +/or SBA	some	Ishii et al. (2005); Lallemand et al. (2005)
Mttp	Signaling	EX	all	Raabe et al. (1998)
Ndst1	Signaling	EX, SBA	5% EX, 5% SBA	Pallerla et al. (2007)
Neurog2	Signaling	EX	5%	Fode et al. (1998); Ma et al. (1998)
Nf1	Signaling	EX	15% females; 0% males	Lakkis et al. (1999)
Nog	Signaling	EX	0-100% EX	McMahon et al. (1998); Stottmann et al. (2006)
*Nuak1,Nuak2** (digenic)*	Signaling	EX + SF + SBA	100%	Ohmura et al. (2012)
*Nuak2**	Signaling	EX	40%	Ohmura et al. (2012)
Numb	Signaling	EX (die by GD11)	all	Zhong et al. (2000); Zilian et al. (2001)
Ovol2	Signaling	No closure	all	Mackay et al. (2006)
p38IP (drey ENU mutant)	Signaling	EX, SBA	55% EX, 15% SBA	Zohn et al. (2006)
*Patz1**	Signaling	EX	25%	Valentino et al. (2013)
Pax2	Signaling	EX	100%	Torres et al. (1996); Puschel et al. (1992)
Pax3 (Sp mutant)	Signaling	EX +/or SBA	50-100% EX; 25-100% SBA	Auerbach (1954); Goulding et al. (1991); Machado et al. (2001)
Pax3 (Sp2H X-ray mutant)	Signaling	EX +/or SBA	35-75% EX, 85% SBA	Fleming & Copp (2000); Machado et al. (2001)
Pax3 (Spd mutant, hypomorph)	Signaling	EX +/or SBA	0-25% EX; 60-100% SBA	Moase & Trasler (1989); Machado et al. (2001)
Pitx2	Signaling	EX	5%	Gage et al. (1999)
Prkaca, Prkacb (digenic)	Signaling	EX	25% in -/-; +/-	Huang et al. (2002)
Ptch1 (partial rescue by Ptch1 transgene, strong MT promoter)	Signaling	EX	some	Milenkovic et al. (1999); Ybot-Gonzalez et al. (2002)
Ptch1 (partial rescue by Ptch1 transgene, weak MT promoter)	Signaling	EX+CRS	all	Milenkovic et al. (1999)
Ptch1 null	Signaling	No closure	all	Goodrich et al. (1997)
Ptch-/- and Cdon-/-	Signaling	Ex	all	Camp et al. (2014)
Ptch-/-, Cdon-/- and Boc-/-	Signaling	Ex	all	Camp et al. (2014)
Ptpn11 chimera	Signaling	SBA	60%	Saxton & Pawson (1999); Feng et al. (1993)
Rac1-/-, Grhl3-Cre/+ (digenic) (Rac1 is conditional null in expression domain of Grhl3)	Signaling	EX +/or SBA	60% EX, 85% SBA	Camerer et al. (2010); Sugihara et al. (1998)
Rara, Rarg (digenic)	Signaling	EX + SF	45%	Lohnes et al. (1994); Ang et al. (1997)
Rbpsuh	Signaling	EX	most	Oka et al. (1995)
Rere (Atr2, om mutant)	Signaling	EX	100%	Zoltewicz et al. (2004)

Rybp heterozygotes	Signaling	EX	15% in heterozygotes	Pirity et al. (2005)
Sall1 truncated	Signaling	EX	40%	Kiefer et al. (2003)
Sall2	Signaling	EX	10% on 129; 0% on C57BL/6	Bohm et al. (2008)
Sall4 heterozygotes	Signaling	EX	5% in heterozygotes	Sakaki-Yumoto et al. (2006)
*Scarb1***	Signaling	EX	44% in females, 20% in males	Santander et al. (2012)
Sfrp1,Sfrp2 (digenic)	Signaling	hydromelia (SB covered by nonneural ectoderm)	90%	Misra & Matise (2010)
Ski	Signaling	EX	5-85% EX	Berk et al. (1997); Colmenares et al. (2002); Lyons et al. (1994)
Smad5	Signaling	EX	some	Chang et al. (1999)
Sp8 (lgl mutant, hypomorph)	Signaling	EX	5%	McNeish et al. (1990); Bell et al. (2003)
Sp8 null	Signaling	EX +/- SF, + SBA	90% EX (some SF), 100% SBA	Bell et al. (2003)
Spint2 (HA12)	Signaling	EX, SBA	95% EX, 10% SBA	Szabo et al. (2009)
Ss18 (Syt)	Signaling	open neural tube, EX	50%	Kimura et al. (2009)
Sufu	Signaling	No closure	all	Cooper et al. (2005); Svard et al. (2006);
T (Tc/tw5 mutant)	Signaling	SBA	100%	Park (1989); Greene et al. (1998)
Tcfap2a (Ap2)	Signaling	SF+EX	100%	Schorle et al. (1996); Zhang et al. (1996); Kohlbecker et al. (2002)
Tcof1+/-	Signaling	EX	5-100% in heterozygotes	Dixon & Dixon (2004)
Tead2	Signaling	EX	30% in -/- dam, 10% in +/- dam	Kaneko et al. (2007)
Tgif1 (exon 3 deletion, truncated)	Signaling	EX	2% in F1 of 129/SvxC57BL/6; 20% in BC1 to C57BL/6	Jin et al. (2006); Kuang et al. (2006)
*Tiam1***	Signaling	EX	30%	Yoo et al. (2012)
Trp53 (p53)	Signaling	EX	20-30% females; 0-1% males	Armstrong et al. (1995); Sah et al. (1995)
p53-/-;Bim+/- and p53-/-;Bim-/-	Signaling	EX and few SBA	100% in female	Delbridge et al. (2019)
Trp53bp2Δ3/Δ3	Signaling	CRS or EX or SBA	8%-46% in different genetic background	Zak et al. (2016)
Tulp3	Signaling	EX +/or SBA	70% EX; 40% SBA	Ikeda et al. (2001)
Tulp3(hhkr mutant) truncated, hypomorph	Signaling	EX +/or SBA	35% EX, 65% SBA	Patterson et al. (2009)
Twist1	Signaling	EX	all	Chen & Behringer (1995); Soo et al. (2002); Bildsoe et al. (2013)
Wnt3a (vt mutant, hypomorph)	Signaling	SBA	some vt/null	Greco et al. (1996)
Ybx1	Signaling	EX	30%	Lu et al. (2005)
Yyl heterozygotes	Signaling	EX	15% in heterozygotes	Donohoe et al. (1999)
Zfhx1a	Signaling	EX or SBA	4% EX, 2% SBA	Takagi et al. (1998)
Zfp36l1	Signaling	EX	some	Stumpo et al. (2004)
Zfp568 (chato mutant)	Signaling	open neural tube	100%	Garcia-Garcia et al. (2008)
Zic2 hypomorph	Signaling	SBA +/- EX	100% SBA, 10% EX	Nagai et al. (1997, 2000)
Zic2Ku (ENU mutant, null)	Signaling	EX (+SBA?)	100% EX (+SBA)?	Elms et al. (2003)
Zic3 (bn mutant)	Signaling	EX	25%	Carrel et al. (2000); Klootwijk et al. (2000); Purandare et al. (2002); Lickert et al. (2005)
Zic5	Signaling	EX	15%	Inoue et al. (2004)
*Alx3***	Transcription & translation	EX (+/-SF)	60% EX, some SF	Lakhwani et al. (2010)
Arnt	Transcription & translation	SF, EX, SF+EX	40%	Kozak et al. (1997)
Brca1 hypomorph	Transcription & translation	EX	10%	Xu et al. (2001)
Brca1 null	Transcription & translation	EX +/or SBA	40%	Gowen et al. (1996)
Cart1	Transcription & translation	EX	65-100%	Zhao et al. (1996)
Cited2	Transcription & translation	EX	60-80%	Dunwoodie et al. (1998); Bamforth et al. (2001); Barbera et al. (2002)
Crebbp	Transcription & translation	EX	all	Tao et al. (1998)
Csnk2a1	Transcription & translation	EX	>90%	Lou et al. (2008); Seldin et al. (2008)
Deaf1	Transcription & translation	EX	80%	Hahm et al. (2004)
Dlx5	Transcription & translation	EX	30%	Depew et al. (1999)
Dnajb6 (Mrj)	Transcription & translation	EX	25%	Watson et al. (2009)
Eif2c2 (Ago2)	Transcription & translation	EX	50%	Liu et al. (2004)
Ep300	Transcription & translation	EX	100%	Yao et al. (1998)
Foxb1	Transcription & translation	EX	5%	Labosky et al. (1997)
Gli3 (Xt, Bph, Pdn mutants)	Transcription & translation	EX	20-60%	Johnson (1969); Hui & Joyner (1993); Sasaki et al. (1997)
*Grhl2 (Axd mutant, hypermorph)***	Transcription & translation	SBA	40-100% (F:M=1:1)	Brouns et al. (2011); Essien et al. (1990)
*Grhl2 (null)***	Transcription & translation	EX+SF, SBA	100% EX+SF, 90% SBA (15% EX+SF in heterozygote)	Brouns et al. (2011); Rifat et al. (2010); Werth et al. (2010)
*Grhl21Nis (ENU mutant, truncated)***	Transcription & translation	EX+SF	100%	Pyrgaki et al. (2011)

Grhl3 null	Transcription & translation	SBA +/- EX	100% SBA, 2% EX	Ting et al. (2003); Auden et al. (2006)
Gtf2i	Transcription & translation	EX	60% in null, 15% in +/-	Enkhmandakh et al. (2009)
Gtf2ird1	Transcription & translation	EX	10% in null, 2% in +/-	Enkhmandakh et al. (2009)
Hes1	Transcription & translation	SF+EX	70%	Ishibashi et al. (1995); Lee et al. (2005a)
Hic1	Transcription & translation	EX	35%	Carter et al. (2000)
Hif1a	Transcription & translation	EX	all	Iyer et al. (1998)
Hipk1, Hipk2 (digenic)	Transcription & translation	EX (F:M=2:1)	50% -/-,-/-; 10%+/-,-/-; 5%-/-,+/-	Isono et al. (2006); Aikawa et al. (2006)
Hmx1 (dmbo mutant)	Transcription & translation	EX	25%	Munroe et al. (2009)
Jarid2 (jmj mutant)	Transcription & translation	EX	50%	Takeuchi et al. (1995)
Lmo4	Transcription & translation	EX	10-50%	Hahm et al. (2004); Tse et al. (2004); Lee et al. (2005b)
Luzp1	Transcription & translation	EX	40%	Hsu et al. (2008)
Med12	Transcription & translation	EX or SBA or CRS	female mosaic hemizygotes: 5% EX, 65% EX+SBA, 30% CRS	Rocha et al. (2010)
Men1	Transcription & translation	EX	20% on 129, 0% on C57BL/6	Bertolino et al. (2003); Lemos et al. (2009)
Msgn1	Transcription & translation	SBA	all?	Yoon & Wold (2000)
Msx1, Msx2 (digenic)	Transcription & translation	EX +/or SBA	some	Ishii et al. (2005); Lallemand et al. (2005)
Neurog2	Transcription & translation	EX	5%	Fode et al. (1998); Ma et al. (1998)
Nup50	Transcription & translation	EX	100%	Smitherman et al. (2000)
*Patz1***	Transcription & translation	EX	25%	Valentino et al. (2013)
Pax2	Transcription & translation	EX	100%	Torres et al. (1996); Puschel et al. (1992)
Pax3 (Sp mutant)	Transcription & translation	EX +/or SBA	50-100% EX; 25-100% SBA	Auerbach (1954); Goulding et al. (1991); Machado et al. (2001)
Pax3 (Sp2H X-ray mutant)	Transcription & translation	EX +/or SBA	35-75% EX, 85% SBA	Fleming & Copp (2000); Machado et al. (2001)
Pax3 (Spd mutant, hypomorph)	Transcription & translation	EX +/or SBA	0-25% EX; 60-100% SBA	Moase & Trasler (1989); Machado et al. (2001)
Pitx2	Transcription & translation	EX	5%	Gage et al. (1999)
Rbpsuh	Transcription & translation	EX	most	Oka et al. (1995)
Rere (Atr2, om mutant)	Transcription & translation	EX	100%	Zoltewicz et al. (2004)
Rpl24 (Bst mutant)	Transcription & translation	EX	30% in Bst/+ (Bst/Bst die in utero)	Rice et al. (1995); Oliver et al. (2004)
Rybp heterozygotes	Transcription & translation	EX	15% in heterozygotes	Pirity et al. (2005)
Sall1 truncated	Transcription & translation	EX	40%	Kiefer et al. (2003)
Sall2	Transcription & translation	EX	10% on 129; 0% on C57BL/6	Bohm et al. (2008)
Sall4 heterozygotes	Transcription & translation	EX	5% in heterozygotes	Sakaki-Yumoto et al. (2006)
Ski	Transcription & translation	EX	5-85% EX	Berk et al. (1997); Colmenares et al. (2002); Lyons et al. (1994)
Sp8 (lgl mutant, hypomorph)	Transcription & translation	EX	5%	McNeish et al. (1990); Bell et al. (2003)
Sp8 null	Transcription & translation	EX +/- SF, + SBA	90% EX (some SF), 100% SBA	Bell et al. (2003)
Ss18 (Syt)	Transcription & translation	open neural tube, EX	50%	Kimura et al. (2009)
T (Tc/tw5 mutant)	Transcription & translation	SBA	100%	Park (1989); Greene et al. (1998)
Tcfap2a (Ap2)	Transcription & translation	SF+EX	100%	Schorle et al. (1996); Zhang et al. (1996); Kohlbecker et al. (2002)
Tcof1+/-	Transcription & translation	EX	5-100% in heterozygotes	Dixon & Dixon (2004)
Tead2	Transcription & translation	EX	30% in -/- dam, 10% in +/- dam	Kaneko et al. (2007)
Trim71 (Mlin41)	Transcription & translation	EX	100%	Maller Schulman et al. (2008)
Trim71 (truncated, hypomorph)	Transcription & translation	EX	55%	Maller Schulman et al. (2008)
Trp53 (p53)	Transcription & translation	EX	20-30% females; 0-1% males	Armstrong et al. (1995); Sah et al. (1995)
p53-/-;Bim+/- and p53-/-;Bim-/-	Transcription & translation	EX and few SBA	100% in female	Delbridge et al. (2019)
Twist1	Transcription & translation	EX	all	Chen & Behringer (1995); Soo et al. (2002); Bildsoe et al. (2013)
Ybx1	Transcription & translation	EX	30%	Lu et al. (2005)
Yyl heterozygotes	Transcription & translation	EX	15% in heterozygotes	Donohoe et al. (1999)
Zfhx1a	Transcription & translation	EX or SBA	4% EX, 2% SBA	Takagi et al. (1998)
Zfp36l1	Transcription & translation	EX	some	Stumpo et al. (2004)
Zfp568 (chato mutant)	Transcription & translation	open neural tube	100%	Garcia-Garcia et al. (2008)
Zic2 hypomorph	Transcription & translation	SBA +/- EX	100% SBA, 10% EX	Nagai et al. (1997, 2000)
Zic2(m1Nisw)				Ying Paper
Zic2Ku (ENU mutant, null)	Transcription & translation	EX (+SBA?)	100% EX (+SBA)?	Elms et al. (2003)
Zic3 (bn mutant)	Transcription & translation	EX	25%	Carrel et al. (2000); Klootwijk et al. (2000); Purandare et al. (2002); Lickert et al. (2005)
Zic5	Transcription & translation	EX	15%	Inoue et al. (2004)
Grhl2 (ENU)	Unknown Function	EX	no data	Zohn et al. (2005)
aln (ENU mutant)	Unknown Function	EX or SBA	low frequency	Herron et al. (2002)

ct mutant (Grhl3 hypomorph?)	Unknown Function	SBA +/- EX	10-20% SBA (1:1=female:male); 0-4% EX (4:1=female:male)	van Straaten & Copp (2001); Greene & Copp (1997); Cogram et al. (2002)
Dbf mutant	Unknown Function	SF or EX+SF	80% SF, 20% EX+SF	Lyon et al. (1996)
Ds mutant	Unknown Function	EX or SBA	0-55% EX, 0-5% SBA	Hummel (1958); Crosby et al. (1992)
excp (ENU mutant)	Unknown Function	EX	25%	Herron et al. (2002)
Exen1, Exen2, Exen3, Exen4 (SELH/Bc strain)	Unknown Function	EX or cleft cerebellum	5-30% EX depending on maternal diet (2:1=female:male); 5-10% cleft cerebellum	Juriloff et al. (1989, 2001, 2006); Harris & Juriloff (2005)
exma mutant	Unknown Function	EX or SF+EX	85% EX, 5% SF+EX	Cunningham et al. (2002)
Gw (ENU mutant)	Unknown Function	EX	no data	Bogani et al. (2004)
l11Jus15 (ENU)	Unknown Function	EX	10%	Kile et al. (2003)
*Line-3P (ENU)***	Unknown Function	EX	30%	Marean et al. (2011)
Mtu (ENU mutant)	Unknown Function	EX	no data	Bogani et al. (2004)
nt mutant	Unknown Function	EX	some	Melloy et al. (1998)
sobp ENU mutant	Unknown Function	EX	no data	Garcia-Garcia et al. (2005)
Thl1-Ex1, Thl1-Ex2 in haplolethal deletions	Unknown Function	EX	50% (hemizygotes)	Howell et al. (2005)
Ts mutant heterozygotes	Unknown Function	EX	5-20% in Ts/+	Ishijima et al. (1998)
*tuft mutant***	Unknown Function	EX or cephalocele	10% EX; 10% cephalocele	Fong et al. (2014)