Additional file 3. Posterior Vertebral Body Fusion Mouse Mutants

Mutant	Affected Vertebrae	Dec. # cdl v	Fused Ribs	Fused Digits	Dev. Structure Affected			Viable	Fertile	Relevant Functions and Pathways
					Somite	N.Tube	N-chord			
am amputated possible mutation: Foxc2 Spontaneous, recessive	data not shown	+++	✓		✓			—		Foxc2 is involved in somite formation; tied to Shh & Bmp [1]
Ankrd13a Gene trapped	cdl							✓	✓	cell-cell adhesion, also involved in neural crest [2]
Arntl (Bmal1 or MOP3) Targeted knock-out	T, S							✓*	✓	circadian clock gene tied to Notch-reg. angiogenesis [3]
Bmp7 Random gene disruption by insertion	T		✓					—		specifies ventroposterior fates and paraxial mesoderm [4]
Cdx2$^{+/-}$*/Cdx4*$^{-/-}$ Targeted knock-outs	S	+++			✓	✓		✓	✓	Hox pathway genes involved in posterior axial elongation; tied to Wnt and Fgf [5]
Cenpj Targeted insertion	cdl			✓				✓	✓	involved in centriole bio-genesis & chrom. stability [6]
CREB Dominant negative knock-in	C, T, L, cdl		✓		✓			—		Modulator of Notch/Wnt in somite segmentation & polarity establishment [7]
Dkk1$^{d/-}$ doubleridge/null compound	L, S, cdl			✓	✓			✓	✓	Lrp6 binding partner; modulates Wnt expression [8]
Dll3 Spontaneous or radiation-induced; recessive	C, T, L, S, cdl	++	✓		✓			✓	✓	Notch/Wnt crosstalker during somitogenesis [9]
Fgf3 Targeted knock-out	cdl	++			✓	✓		✓*	✓	Upstream modulator of Notch/Wnt during somite polarity establishment [10]
Fkbp8 Gene trapped	T, L, S					✓		✓	✓	Neural tube patterning; tied to Notch via Zic1 down-reg. [11]

Gnai3 Targeted knock-out	L	data not shown	✓		✓			✓	✓	tied to Bmp7, and Wnt/axin signaling [12]
Hes7 Targeted insertion knock-in	C, T, L, S, cdl	++	✓		✓			✓*	✓	Notch/Fgf crosstalker in somite segmentation clock [13]
Ikkα Targeted reporter insertion	C, S, cdl	+		✓				—		involved in epidermal cell-cell adhesion; facilitates bone formation via bFGF [14]
Jsr jumbled spine & ribs Spontaneous, dominant	T, cdl	++	✓		✓			✓	✓	mutation unknown; tied to Notch pathway via Uncx [15]
Knk kinked tail Spontaneous, semi-dominant	cdl	+					✓	✓	✓	mutation unknown [16]
Lfng Chemically induced single point mutation	T, L, S	++	✓		✓			✓*	✓	Notch pathway; involved in somite segmentation clock [17]
Lrp6 Spontaneous, semi-dominant	cdl	++	✓		✓	✓		✓*	✓	Wnt/Notch crosstalker in somite segmentation [18]
mea meander tail Spontaneous, recessive	T, L, cdl + S vertebral spines	++						✓	✓	mutation unknown [19]
Meox1/Meox2 Targeted knock-outs	cdl	++	✓		✓			—		involved in somite patterning; induced by retinoic acid [20]
Mesp2 Targeted insertion knock-out	C; L transverse process fusion	++			✓			✓*	✓	Notch pathway; involved in somite boundary formation [21]
N-myc alternative splice conditional mutation	L						✓	✓	✓	promotes neurogenesis via N-myc/Dll3/Notch cascade [22]
Noto Spontaneous single point mutation, recessive	L, S, cdl	++			✓	✓	✓	✓	✓	involved in notochord extension & node; controlled by Wnt [23]
Nrarp Targeted knock-out	T, L, S, cdl	+	✓		✓			✓	✓	Notch/Wnt crosstalker in somite segmentation clock [24]
Pax3 splotch[d] Spontaneous, semi-dominant	C, T, L, S		✓		✓	✓		—		involved in preaxial meso-derm condensation for somitogenesis; tied toNotch pathway genes [25]
Plxnd1 Targeted knock-out	T, L	data not shown	✓					—		Notch-controlled role in inter-somitic vascular development [26]

Ppp5c Targeted insertion	cdl							✓	✓	regulates ERK signaling [27]; ERK signaling is involved in Wnt/FGF controlled somite segmentation [28]
rh rachiterata Spontaneous, recessive	L, S	+	✓		✓			✓	✓	mutation unknown; tied to Shh via Pax1 [25]
Ror2 Targeted insertion knock-in	cdl	+	✓	✓	✓			✓	✓	Wnt Rc; involved in maintenance of presomitic mesoderm; also tied to Notch via Dll1, Lfng, & Mesp2 down-reg. [29]
Rpl38 Spontaneous, semi-dominant	C, T, cdl	++	✓			✓		✓*	✓	ribosomal protein involved in Hox gene control [30]
Rps7 Chemically induced single point mutation, dominant	cdl; S transverse process fusion	+						✓*	✓	ribosomal protein; role in posterior dev. not studied [31]
Sfxn1 f, flexed tail Spontaneous, recessive	L, S, cdl	+/-	✓			✓	✓	✓	✓	Smad5/Bmp4 signaling defects [32]
Sulf1/Sulf2 Targeted insertion knock-outs	T, L, S, cdl				✓			✓	✓	regulate growth factors; tied to Bmp & Shh signaling [33]
Tbx6 Spontaneous, duplication/ insertion	C; T, L trans-verse process fusion	+	✓		✓	✓	✓	✓	✓*	Notch-controlled role in somite boundary formation [34]
Uncx Targeted reporter insertion	data not shown		✓		✓			—		Notch signaling modulator in somite condensation and differentiation [35]
Vangl2 (homozygote) Spontaneous, single point mutation, semi-dominant	T, L, S, cdl	++	✓			✓		—		involved in convergent extension & cell polarity; tied to Wnt [36]
Wnt5a Targeted insertion knock-out	S, cdl	+++			✓	✓		—		Wnt secreted by the VER [37]; strain-dep. vertebral fusion & viability defects