

Mouse phenotype	Wildtype		Nipbl+/-		Assay/ Method	Comparable clinical findings in CdLS	Incidence	Reference
	%	N	%	N				
Prenatal Growth Retardation (E17.5 - E18.5)	0	37	100	36	Weight (Fig. 2)	Low birth weight (< 2500 g)	56-68%	Kline et al., 1993; Selicorni et al., 2007
Skeletal Abnormalities (E18.5) -Ossification delay -Longer olecranon process	0 0	7 7	100 100	7 7	Bone Staining (Fig. 1)	Overall skeletal abnormalities Ulnar dys-/hypoplastic changes	>80% 47-64%	Roposch et al., 2004; Kline et al., 2007b
Atrial septal defect (ASD) (E15.5-E17.5)	0	6	58.3	12	Histology (Fig. 1)	Congenital heart defects; ASD (clinically significant)	26-45% 12-21%	Jackson et al., 1993; Mehta and Ambalavanan, 1997; Tsukahara et al., 1998; Selicorni et al., 2007; Barisic et al., 2008
¹ Postnatal Growth Retardation	0	287	100	173	Weight (Fig. 2)	-Postnatal Growth Retardation (both genders)	63%	Kline et al., 1993; Kline et al., 2007a
Craniofacial dysmorphism	0	40	100	23	Micro-CT (Fig.3)	Craniofacial dysmorphism	100%	Ireland et al., 1993; Allanson et al., 1997; Kline et al., 2007b
Microcephaly	0	40	100	23	Micro-CT (Fig. 4)	-Microcephaly	33-73%	Kline et al., 1993; Kline et al., 2007a; Selicorni et al., 2007; Barisic et al., 2008
^{1,2} Ophthalmic defects (Adult mice only, i.e. 4 weeks of age and older)	1.4	287	22.0	173	Morphology/ Histology (Fig. 4)	-Blepharitis -Microcornea -Nasolacrimal duct obstruction, myopia, ptosis	25% 21% 46-66%	Levin et al., 1990; Wygnanski-Jaffe et al., 2005; Nallasamy et al., 2006
³ Hearing Deficits	6.7	15	92.9	14	ABR (Fig. 4)	-Sensorineural hearing loss -Conductive hearing loss	20-85% 60%	Sataloff et al., 1990; Sakai et al., 2002; Marchisio et al., 2008

¹Postnatal data only reflect incidence among animals that survived to weaning. ²Eye defects include: central corneal opacity, microphthalmia, swelling, and/or closure/sealing of eye. ³Hearing defects include: reduction in peak III of ABR, deafness or increased stimulus threshold for ABR.

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