

Vertebrate Ciliary Mutants

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| Mutant name | Human ortholog | Species | Mutant type | Reference |
|------------------------------------|----------------|--------------------|--------------------------------|---------------------------------|
| <i>Ak7</i> ^{-/-} | AK7 | <i>M. musculus</i> | transgene insertion | Fernandez-Gonzalez et al., 2008 |
| <i>Alms1</i> ^{-/-} | ALMS1 | <i>M. musculus</i> | gene trap | Collin et al., 2005 |
| <i>hennin</i> | ARL13B | <i>M. musculus</i> | ENU | Caspary et al., 2007 |
| <i>hi459/scorpion</i> | ARL13B | <i>D. rerio</i> | retrovirus insertion | Sun et al., 2004 |
| <i>stumpy</i> ^{fl/fl} | B9D2 | <i>M. musculus</i> | targeted conditional knock-out | Town et al., 2008 |
| <i>Bbs1</i> ^{-/-} | BBS1 | <i>M. musculus</i> | gene trap | Kulaga et al., 2004 |
| <i>Bbs1</i> ^{M390R/M390R} | BBS1 | <i>M. musculus</i> | targeted knock-in | Davis et al., 2007 |
| <i>Bbs2</i> ^{-/-} | BBS2 | <i>M. musculus</i> | targeted knock-out | Nishimura et al., 2004 |
| <i>Bbs4</i> ^{-/-} | BBS4 | <i>M. musculus</i> | gene trap, targeted knock-out | Kulaga et al., 2004 |
| <i>Bbs6</i> ^{-/-} | BBS6 | <i>M. musculus</i> | gene trap, targeted knock-out | Ross et al., 2005, |
| <i>bpk</i> | BICC1 | <i>M. musculus</i> | spontaneous | Cogswell et al., 2003 |
| <i>jcpk</i> | BICC1 | <i>M. musculus</i> | chlorambucil mutagenesis | Cogswell et al., 2003 |
| <i>Hty</i> | C2CD3 | <i>M. musculus</i> | ENU? | Hoover et al., 2008 |
| <i>Cby</i> ^{-/-} | CBY1 | <i>M. musculus</i> | targeted knock-out | Voronina et al., 2009 |
| <i>sentinel</i> | CC2D2A | <i>D. rerio</i> | ENU | Gorden et al., 2008 |
| <i>waltzer</i> | CDH23/USH1D | <i>M. musculus</i> | spontaneous | Di Palma et al., 2001 |
| <i>sputnik</i> | CDH23/USH1D | <i>D. rerio</i> | ENU | Sollner et al., 2004 |
| <i>rd16</i> | CEP290/NPHP6 | <i>M. musculus</i> | spontaneous | Chang et al., 2006 |
| <i>rdAc</i> | CEP290/NPHP6 | <i>C. felis</i> | spontaneous | Menotti-Raymond et al., 2007 |
| <i>hi3959A/qilin</i> | CLUAP1 | <i>D. rerio</i> | retrovirus insertion | Sun et al., 2004 |
| <i>rd8</i> | CRB1 | <i>M. musculus</i> | spontaneous | Mehalow et al., 2003 |
| <i>oko meduzy</i> | CRB2 | <i>D. rerio</i> | ENU | Omori et al., 2006 |
| <i>Rootletin</i> ^{-/-} | CROCC | <i>M. musculus</i> | targeted knock-out | Yang et al., 2005 |
| <i>cpk</i> | CYS1 | <i>M. musculus</i> | spontaneous | Hou et al., 2002 |
| <i>mikre oko</i> | DCTN1 | <i>D. rerio</i> | ENU | Tsujikawa et al., 2007 |
| <i>whirler</i> | DFNB31 | <i>M. musculus</i> | spontaneous | Mburu et al., 2003 |
| <i>Mdhc7</i> ^{-/-} | DNAH1 | <i>M. musculus</i> | targeted knock-out | Neesen et al., 2001 |
| <i>iv</i> | DNAHC11 | <i>M. musculus</i> | spontaneous | Supp et al., 1997 |
| <i>lgl</i> | DNAHC11 | <i>M. musculus</i> | random insertion | Supp et al., 1997 |
| <i>Dnahc5</i> ^{del593} | DNAHC5 | <i>M. musculus</i> | ENU | Tan et al., 2007 |
| <i>Mdnah5</i> ^{-/-} | DNAHC5 | <i>M. musculus</i> | random insertion | Ibañez-Tallon et al., 2002 |
| <i>ling-ling/Dnchc2</i> | DYNC2H1 | <i>M. musculus</i> | ENU | Huangfu et al., 2005 |
| <i>E2f4</i> ^{-/-} | E2F4 | <i>M. musculus</i> | targeted knock-out | Danielian et al., 2007 |
| <i>Efhc1</i> ^{-/-} | EFHC1 | <i>M. musculus</i> | targeted knock-out | Suzuki et al., 2009 |

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|---|---------------|----------------------|--------------------------------|---|
| <i>Evc</i> ^{-/-} | EVC | <i>M. musculus</i> | targeted knock-out | Ruiz-Perez et al., 2007 |
| <i>Fat4</i> ^{-/-} | FAT4 | <i>M. musculus</i> | targeted conditional knock-out | Saburi et al., 2008 |
| <i>Hfh-4</i> ^{-/-} | FOXJ1 | <i>M. musculus</i> | targeted knock-out | Chen et al., 1998 |
| <i>Glis2</i> ^{lacZ/lacZ} | GLIS2/NPHP7 | <i>M. musculus</i> | targeted knock-out | Attanasio et al., 2007 |
| <i>Glis3</i> ^{zf/zf} | GLIS3 | <i>M. musculus</i> | targeted knock-out | Kang et al., 2009 |
| <i>Vlgr1</i> ^{del7TM/del7TM} | GPR98 | <i>M. musculus</i> | targeted knock-out | McMillan et al., 2004 |
| <i>Hsf1</i> ^{-/-} | HSF1 | <i>M. musculus</i> | targeted knock-out | Takaki et al., 2007 |
| <i>hy3</i> | HYDIN | <i>M. musculus</i> | spontaneous, random insertion | Robinson et al., 2003, Davy et al., 2003 |
| <i>alien</i> | IFT139/TTC21B | <i>M. musculus</i> | ENU | Tran et al., 2008 |
| <i>Med1</i> ^{Δ1-3} | IFT122/WDR10 | <i>M. musculus</i> | targeted knock-out of Med1 | Cortellino et al., 2008 |
| <i>hi2211/moe</i> | IFT172 | <i>D. rerio</i> | retrovirus insertion | Sun et al., 2004 |
| <i>lft172</i> ^{-/-} | IFT172 | <i>M. musculus</i> | targeted knock-out | Gorivodsky et al., 2008 |
| <i>wimple</i> | IFT172 | <i>M. musculus</i> | ENU | Huangfu et al., 2003 |
| <i>lft20</i> ^{flox/flox} | IFT20 | <i>M. musculus</i> | targeted conditional knock-out | Jonassen et al., 2008 |
| <i>hi3417/curly</i> | IFT57 | <i>D. rerio</i> | retrovirus insertion | Sun et al., 2004 |
| <i>Hippi</i> ^{-/-} | IFT57 | <i>M. musculus</i> | targeted knock-out | Houde et al., 2006 |
| <i>hi409/larry</i> | IFT81 | <i>D. rerio</i> | retrovirus insertion | Sun et al., 2004 |
| <i>oval</i> | IFT88 | <i>D. rerio</i> | ENU | Tsujikawa et al., 2004 |
| <i>flexo</i> | IFT88 | <i>M. musculus</i> | ENU | Huangfu et al., 2003 |
| <i>Tg737</i> ^{orpk} / <i>polaris</i> | IFT88 | <i>M. musculus</i> | random insertion | Moyer et al., 1994, Pazour et al., 2000 |
| <i>lft88</i> ^{fl/fl} | IFT88 | <i>M. musculus</i> | targeted conditional knock-out | Haycraft et al., 2007 |
| <i>cobblestone</i> | IFT88 | <i>M. musculus</i> | ENU | Willaredt et al., 2008 |
| <i>inv</i> | INVS/NPHP2 | <i>M. musculus</i> | random insertion | Mochizuki et al., 1998 |
| <i>Kif3a</i> ^{-/-} | KIF3A | <i>M. musculus</i> | targeted knock-out | Marszalek et al., 1999, Takeda et al., 1999 |
| <i>Kif3b</i> ^{-/-} | KIF3B | <i>M. musculus</i> | targeted knock-out | Nonaka et al., 1998 |
| <i>ktu</i> | KTU | <i>O. latipes</i> | ENU | Omran et al., 2008 |
| <i>seahorse</i> | LRRC6 | <i>D. rerio</i> | retrovirus insertion | Kishimoto et al., 2008 |
| <i>hu255H</i> | LRRC50 | <i>D. rerio</i> | ENU | van Rooijen et al., 2008 |
| <i>swt</i> | LRRC50 | <i>D. rerio</i> | ENU | Sullivan-Brown et al., 2007 |
| <i>mindbomb</i> | MIB1 | <i>D. rerio</i> | ENU | Ma et al., 2007, Liu et al., 2007 |
| <i>wpk</i> | MKS3 | <i>R. norvegicus</i> | spontaneous | Smith et al., 2006 |
| <i>bpck</i> | MKS3 | <i>M. musculus</i> | spontaneous | Cook et al., 2009 |
| <i>mariner</i> | MYO7A | <i>D. rerio</i> | ENU | Ernest et al., 2000 |
| <i>shaker1</i> | MYO7A | <i>M. musculus</i> | spontaneous, ENU | Self et al., 1998 |
| <i>kat</i> | NEK1 | <i>M. musculus</i> | spontaneous | Upadhya et al., 2000 |
| <i>jck</i> | NEK8 | <i>M. musculus</i> | spontaneous | Liu et al., 2002 |
| <i>Noto</i> ^{GFP/GFP} | NOTO | <i>M. musculus</i> | targeted knock-out | Beckers et al., 2007 |
| <i>Nphp1</i> ^{-/-} | NPHP1 | <i>M. musculus</i> | targeted knock-out | Jiang et al., 2009 |
| <i>pcy</i> | NPHP3 | <i>M. musculus</i> | spontaneous | Omran et al., 2001 |
| <i>Nphp3</i> ^{-/-} | NPHP3 | <i>M. musculus</i> | targeted knock-out | Bergmann et al., 2008 |

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|---|-----------------|----------------------|--------------------------------|---------------------------------------|
| <i>crdSWHD</i> | <i>NPHP4</i> | <i>C. familiaris</i> | spontaneous | Wiik et al. 2008 |
| <i>Ofd1</i> ^{-/-} | <i>OFD1</i> | <i>M. musculus</i> | targeted knock-out | Ferrante et al., 2006 |
| <i>Lis1</i> ^{-/-} | <i>PAFAH1B1</i> | <i>M. musculus</i> | targeted knock-out | Hirotsune et al., 1998 |
| <i>Par3</i> ^{ΔE3} | <i>PAR3</i> | <i>M. musculus</i> | targeted knock-out | Fan et al., 2004, Hirose et al., 2006 |
| <i>Ames waltzer</i> | <i>PCDH15</i> | <i>M. musculus</i> | spontaneous | Alagramam et al., 2001 |
| <i>orbiter</i> | <i>PCDH15</i> | <i>M. musculus</i> | ENU | Seiler et al., 2005 |
| <i>Pkd1</i> ^{-/-} | <i>PKD1</i> | <i>M. musculus</i> | targeted knock-out | Lu et al., 1997 |
| <i>hi4166</i> | <i>PKD2</i> | <i>D. rerio</i> | retrovirus insertion | Sun et al., 2004 |
| <i>cup</i> | <i>PKD2</i> | <i>D. rerio</i> | ENU | Schottenfeld et al., 2007 |
| <i>Pkd2</i> ^{-/-} | <i>PKD2</i> | <i>M. musculus</i> | targeted knock-out | Wu et al., 1998 |
| <i>pck</i> | <i>PKHD1</i> | <i>R. norvegicus</i> | spontaneous | Ward et al., 2002 |
| <i>Pkhd1</i> ^{flox3-4/flox3-4} | <i>PKHD1</i> | <i>M. musculus</i> | targeted conditional knock-out | Garcia-Gonzalez et al., 2007 |
| <i>talpid</i> ³ | <i>PPL</i> | <i>G. domesticus</i> | spontaneous | Yin et al., 2009 |
| <i>Rfx3</i> ^{flox/flox} | <i>RFX3</i> | <i>M. musculus</i> | targeted conditional knock-out | Bonnafe et al., 2004 |
| <i>Rpgr</i> ^{-/-} | <i>RPGR</i> | <i>M. musculus</i> | targeted knock-out | Hong et al., 2000 |
| <i>Rpgrip1</i> ^{-/-} | <i>RPGRIP1</i> | <i>M. musculus</i> | targeted insertion | Zhao et al., 2003 |
| <i>crdMLHD</i> | <i>RPGRIP1</i> | <i>C. familiaris</i> | spontaneous | Mellersh et al., 2006 |
| <i>Ftm</i> ^{-/-} | <i>RPGRIP1L</i> | <i>M. musculus</i> | targeted knock-out | Vierkotten et al., 2007 |
| <i>Spag16L</i> ^{-/-} | <i>SPAG16</i> | <i>M. musculus</i> | targeted knock-out | Zhang et al., 2006 |
| <i>Spag6</i> ^{-/-} | <i>SPAG6</i> | <i>M. musculus</i> | targeted knock-out | Sapiro et al., 2002 |
| <i>nm1054</i> | <i>STEAP3</i> | <i>M. musculus</i> | spontaneous | Lee et al., 2008 |
| <i>Fu</i> ^{-/-} | <i>STK36</i> | <i>M. musculus</i> | targeted knock-out | Wilson et al., 2009 |
| <i>Tektin-t</i> ^{-/-} | <i>TEKT2</i> | <i>M. musculus</i> | gene trap | Tanaka et al., 2004 |
| <i>elipsa</i> | <i>TRAF3IP1</i> | <i>D. rerio</i> | ENU | Omori et al., 2008 |
| <i>Gmap210</i> ^{-/-} | <i>TRIP11</i> | <i>M. musculus</i> | <i>targeted knock-out</i> | Follit et al., 2008 |
| <i>Tsc1</i> ^{-/-} | <i>TSC1</i> | <i>M. musculus</i> | targeted knock-out | Hartman et al. 2009 |
| <i>Tsc1</i> ^{-/-} | <i>TSC2</i> | <i>M. musculus</i> | targeted knock-out | Hartman et al. 2009 |
| <i>fleer</i> | <i>TTC30B</i> | <i>D. rerio</i> | ENU | Pathak et al., 2008 |
| <i>deaf circler</i> | <i>USH1C</i> | <i>M. musculus</i> | spontaneous | Johnson et al., 2003 |
| <i>Jackson shaker</i> | <i>USH1G</i> | <i>M. musculus</i> | spontaneous | Weil et al., 2003 |
| <i>Ush2a</i> ^{-/-} | <i>USH2A</i> | <i>M. musculus</i> | targeted knock-out | Liu et al., 2007 |
| <i>Ltap</i> | <i>VANGL2</i> | <i>M. musculus</i> | spontaneous | Ross et al., 2005 |
| <i>strabismus/trilobite</i> | <i>VANGL2</i> | <i>D. rerio</i> | ENU | Ross et al., 2005 |

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