

TABLE 1. Comprehensive list of ciliary genes associated with retinal dystrophies in syndromic and non-syndromic disorders^{a,b,c}.

| | Non-syndromic retinal ciliopathies | | | | Syndromic ciliopathies with retinal degeneration | | | | | | | | | | | | | | Other ciliopathies without retinal degeneration | | | | | References | |
|---------------|------------------------------------|------|-----|----|--|-----|-----|------|-------|------|-------|-----|------|------|------|-----|------|-----|---|------|------|------|-----|------------|--|
| | RP | CORD | LCA | MD | AS | BBS | CED | CILD | CRDHL | JBTS | MCCRP | MKS | MORM | MRC5 | OFDS | SLS | SRTD | USH | COACH | LGMD | MKKS | NPHP | NSD | | SGBS |
| <i>AHI1</i> | ✓ | | | | | | | | | ✓ | | | | | | | | | | | | | | | (Nguyen et al., 2017) |
| <i>AIPL1</i> | | ✓ | ✓ | | | | | | | | | | | | | | | | | | | | | | (Sacristan-Reviriego et al., 2020) |
| <i>ALMS1</i> | | ✓ | ✓ | | ✓ | | | | | | | | | | | | | | | | | | | | (Xu et al., 2016) |
| <i>ARL13B</i> | | | | | | | | | | ✓ | | | | | | | | | | | | | | | (Cantagrel et al., 2008) |
| <i>ARL2</i> | | | | | | | | | | | | | | ✓ | | | | | | | | | | | (Cai et al., 2019) |
| <i>ARL2BP</i> | ✓ | | | | | | | | | | | | | | | | | | | | | | | | (Davidson et al., 2013) |
| <i>ARL3</i> | ✓ | ✓ | | | | | | | | ✓ | | | | | | | | | | | | | | | (Sheikh et al., 2019) |
| <i>ARL6</i> | ✓ | | | | | ✓ | | | | | | | | | | | | | | | | | | | (Zenteno et al., 2020) |
| <i>ARMC9</i> | | | | | | | | | | ✓ | | | | | | | | | | | | | | | (Van De Weghe et al., 2017) |
| <i>ARSG</i> | | | | | | | | | | | | | | | | | | ✓ | | | | | | | (Abad-Morales et al., 2020) |
| <i>B9D1</i> | | | | | | | | | | ✓ | | ✓ | | | | | | | | | | | | | (Romani et al., 2014) |
| <i>B9D2</i> | | | | | | | | | | ✓ | | ✓ | | | | | | | | | | | | | (Radhakrishnan et al., 2019) |
| <i>BBS1</i> | ✓ | | | | | ✓ | | | | ✓ | | | | | | | | | | | | | | | (Estrada-Cuzcano et al., 2012a; Suzuki et al., 2016) |
| <i>BBS10</i> | | | | | | ✓ | | | | | | | | | | | | | | | | | | | (Suspitsin and Imyanitov, 2016) |
| <i>BBS12</i> | | | | | | ✓ | | | | | | | | | | | | | | | | | | | (Suspitsin and Imyanitov, 2016) |
| <i>BBS18</i> | | | | | | ✓ | | | | | | | | | | | | | | | | | | | (Suspitsin and Imyanitov, 2016) |
| <i>BBS2</i> | ✓ | | | | | ✓ | | | | | | | | | | | | | | | | | | | (Suspitsin and Imyanitov, 2016) |

| | Non-syndromic retinal ciliopathies | | | | Syndromic ciliopathies with retinal degeneration | | | | | | | | | | | | | | Other ciliopathies without retinal degeneration | | | | | | References |
|----------------|------------------------------------|------|-----|----|--|-----|-----|------|-------|------|-------|-----|------|------|------|-----|------|-----|---|------|------|------|-----|------|---|
| | RP | CORD | LCA | MD | AS | BBS | CED | CILD | CRDHL | JBTS | MCCRP | MKS | MORM | MRC5 | OFDS | SLS | SRTD | USH | COACH | LGMD | MKKS | NPHP | NSD | SGBS | |
| <i>BBS4</i> | | | | | | ✓ | | | | | | | | | | | | | | | | | | | (Suspitsin and Imyanitov, 2016)f |
| <i>BBS5</i> | | | | | | ✓ | | | | | | | | | | | | | | | | | | | (Suspitsin and Imyanitov, 2016) |
| <i>BBS7</i> | | | | | | ✓ | | | | | | | | | | | | | | | | | | | (Suspitsin and Imyanitov, 2016) |
| <i>BBS8</i> | ✓ | | | | | ✓ | | | | | | | | | | | | | | | | | | | (Suspitsin and Imyanitov, 2016) |
| <i>BBS9</i> | | | | | | ✓ | | | | | | | | | | | | | | | | | | | (Suspitsin and Imyanitov, 2016) |
| <i>C2orf71</i> | ✓ | ✓ | | | | | | | | | | | | | | | | | | | | | | | (Corral-Serrano et al., 2018) |
| <i>C8orf37</i> | ✓ | ✓ | | | | ✓ | | | | | | | | | | | | | | | | | | | (Tatour and Ben-yosef, 2020) |
| <i>CC2D2A</i> | ✓ | | | | | | | | ✓ | | ✓ | | | | | | | ✓ | | | | | | | (Mjécaese et al., 2019) |
| <i>CDH23</i> | | | | | | | | | | | | | | | | | ✓ | | | | | | ✓ | | (Whatley et al., 2020) |
| <i>CEP104</i> | | | | | | | | | ✓ | | | | | | | | | | | | | | | | (Bachmann-Gagescu et al., 2020) |
| <i>CEP120</i> | | | | | | | | | ✓ | | | | | | | | ✓ | | | | | | | | (Bachmann-Gagescu et al., 2020) |
| <i>CEP164</i> | | | | | | | | | | | | | | | | ✓ | | | | | | ✓ | | | (Chaki et al., 2012) |
| <i>CEP19</i> | | | | | | ✓ | | | | | | | | | | | | | | | | | | | (Kanie et al., 2017) |
| <i>CEP250</i> | ✓ | | | | | | | ✓ | | | | | | | | | | ✓ | | | | | | | (Kubota et al., 2018) |
| <i>CEP290</i> | ✓ | ✓ | ✓ | | | ✓ | | | ✓ | | ✓ | | | | | ✓ | | | | | | ✓ | | | (Rachel et al., 2012; Roosing et al., 2017) |
| <i>CEP41</i> | | | | | | | | | ✓ | | | | | | | | | | | | | | | | (Bachmann-Gagescu et al., 2020) |
| <i>CEP78</i> | | ✓ | | | | | | ✓ | | | | | | | | | | | | | | | | | (Namburi et al., 2016; Gill et al., 2019) |

| | Non-syndromic retinal ciliopathies | | | | Syndromic ciliopathies with retinal degeneration | | | | | | | | | | | | | | | Other ciliopathies without retinal degeneration | | | | | | References |
|----------------|------------------------------------|------|-----|----|--|-----|-----|------|-------|------|-------|-----|------|------|------|-----|------|-----|-------|---|------|------|-----|------|--|------------|
| | RP | CORD | LCA | MD | AS | BBS | CED | CILD | CRDHL | JBTS | MCCRP | MKS | MORM | MRC5 | OFDS | SLS | SRTD | USH | COACH | LGMD | MKKS | NPHP | NSD | SGBS | | |
| <i>CEP83</i> | | | | | | | | | | ✓ | | | | | | | | | | | | ✓ | | | (Braun and Hildebrandt, 2017) | |
| <i>CLRN1</i> | ✓ | | | | | | | | | | | | | | | | | ✓ | | | | | | | (Tatour and Ben-yosef, 2020) | |
| <i>CSPP1</i> | | | | | | | | | | ✓ | | | | | | | | | | | | | | | (Bachmann-Gagescu et al., 2020) | |
| <i>DYNC2H1</i> | ✓ | | | | | | | | | | | | | | | | ✓ | | | | | | | | (Vig et al., 2020) | |
| <i>FAM161A</i> | ✓ | | | | | | | | | | | | | | | | | | | | | | | | (Bachmann-Gagescu and Neuhaus, 2019) | |
| <i>FSCN2</i> | ✓ | | | | | | | | | | | | | | | | | | | | | | | | (Chen et al., 2019) | |
| <i>IFT139</i> | | | | | | | | | | | | | | | | | ✓ | | | | | ✓ | | | (Niwa, 2016) | |
| <i>IFT140</i> | ✓ | | ✓ | | | | | | | | | | | | | | ✓ | | | | | | | | (Xu et al., 2015) | |
| <i>IFT144</i> | | | | | | | ✓ | | | | | | | | ✓ | ✓ | | | | | | ✓ | | | (Braun and Hildebrandt, 2017) | |
| <i>IFT172</i> | ✓ | | | | | ✓ | | | | ✓ | | | | | ✓ | ✓ | | | | | | | | | (Yamada et al., 2019) | |
| <i>IFT27</i> | | | | | | ✓ | | | | | | | | | | | | | | | | | | | (Suspitsin and Imyanitov, 2016) | |
| <i>IFT38</i> | | | ✓ | | | | | | | | | | | | | | | | | | | | | | (Soens et al., 2016) | |
| <i>IFT43</i> | ✓ | | | | | ✓ | | | | | | | | | | | ✓ | | | | | | | | (Biswas et al., 2017) | |
| <i>IFT52</i> | | | | | | ✓ | | | | | | | | | | | ✓ | | | | | | | | (Girisha et al., 2016; Chen et al., 2018) | |
| <i>IFT54</i> | | | | | | ✓ | | | | | | | | | ✓ | ✓ | | | | | | | | | (Bizet et al., 2015; Zhang et al., 2018) | |
| <i>IFT80</i> | | | | | | | | | | | | | | | | | ✓ | | | | | | | | (Zhang et al., 2018) | |
| <i>IFT81</i> | | ✓ | | | | | | | | | | | | | | | ✓ | | | | | | | | (Duran et al., 2016; Dharmat et al., 2017) | |

| | Non-syndromic retinal ciliopathies | | | | Syndromic ciliopathies with retinal degeneration | | | | | | | | | | | | | | | Other ciliopathies without retinal degeneration | | | | | References |
|-----------------|------------------------------------|------|-----|----|--|-----|-----|------|-------|------|-------|-----|------|------|------|-----|------|-----|-------|---|------|------|-----|------|--|
| | RP | CORD | LCA | MD | AS | BBS | CED | CILD | CRDHL | JBTS | MCCRP | MKS | MORM | MRC5 | OFDS | SLS | SRTD | USH | COACH | LGMD | MKKS | NPHP | NSD | SGBS | |
| <i>INPP5E</i> | | | ✓ | | | | | | | ✓ | | | ✓ | | | | | | | | | | | | (Wang and Deretic, 2014; Fansa et al., 2016) |
| <i>INVS</i> | | | | | | | | | | | | | | | | ✓ | | | | | | ✓ | | | (O'Toole et al., 2006) |
| <i>KCNJ13</i> | | | ✓ | | | | | | | | | | | | | | | | | | | | | | (Sergouniotis et al., 2011) |
| <i>KIAA0556</i> | | | | | | | | | | ✓ | | | | | | | | | | | | | | | (Bachmann-Gagescu et al., 2020) |
| <i>KIAA0586</i> | | | | | | | | | | ✓ | | | | | | | ✓ | | | | | | | | (Bachmann-Gagescu et al., 2020) |
| <i>KIAA1549</i> | ✓ | | | | | | | | | | | | | | | | | | | | | | | | (De Bruijn et al., 2018) |
| <i>KIF3B</i> | | | | | | | | | | | | | | | | ✓ | | | | | | | | | (Cogné et al., 2020) |
| <i>KIF7</i> | | | | | | | | | | ✓ | | | | | | | | | | | | | | | (Bachmann-Gagescu et al., 2020) |
| <i>KIZ</i> | ✓ | | | | | | | | | | | | | | | | | | | | | | | | (El Shamieh et al., 2014) |
| <i>LCA5</i> | ✓ | ✓ | ✓ | | | | | | | | | | | | | | | | | | | | | | (Chen et al., 2016) |
| <i>LZTFL1</i> | | | | | | ✓ | | | | | | | | | | | | | | | | | | | (Susptsin and Imyanitov, 2016) |
| <i>MAK</i> | ✓ | | | | | | | | | | | | | | | | | | | | | | | | (Gray et al., 2018) |
| <i>MKKS</i> | | | | | | ✓ | | | | | | | | | | | | | | | ✓ | | | | (Susptsin and Imyanitov, 2016) |
| <i>MKS1</i> | | | | | | ✓ | | | | ✓ | | ✓ | | | | | | | | | | | | | (Bader et al., 2016) |
| <i>MYO7A</i> | | | ✓ | | | | | | | | | | | | | | | ✓ | | | | | ✓ | | (Wang et al., 2011; Whatley et al., 2020) |
| <i>NEK2</i> | ✓ | | | | | | | | | | | | | | | | | | | | | | | | (Nishiguchi et al., 2013) |
| <i>NEK8</i> | | | | | | | | | | ✓ | | ✓ | | | | | | ✓ | | | | ✓ | | | (Ronquillo et al., 2012) |
| <i>NPHP1</i> | | | | | | ✓ | | | | ✓ | | | | | | ✓ | | | | | | ✓ | | | (Susptsin and Imyanitov, 2016) |

| | Non-syndromic retinal ciliopathies | | | | Syndromic ciliopathies with retinal degeneration | | | | | | | | | | | | | | | Other ciliopathies without retinal degeneration | | | | | | References |
|-----------------|------------------------------------|------|-----|----|--|-----|-----|------|-------|------|-------|-----|------|------|------|-----|------|-----|-------|---|------|------|-----|------|--|------------|
| | RP | CORD | LCA | MD | AS | BBS | CED | CILD | CRDHL | JBTS | MCCRP | MKS | MORM | MRC5 | OFDS | SLS | SRTD | USH | COACH | LGMD | MKKS | NPHP | NSD | SGBS | | |
| <i>NPHP3</i> | | | | | | | | | | | | ✓ | | | | ✓ | | | | | | ✓ | | | (Luo and Tao, 2018) | |
| <i>NPHP4</i> | | | | | | | | | | | | | | | | ✓ | | | | | | ✓ | | | (Luo and Tao, 2018) | |
| <i>NPHP5</i> | ✓ | | ✓ | | | | | | | | | | | | | ✓ | | | | | | ✓ | | | (Tong et al., 2013; Luo and Tao, 2018) | |
| <i>OFD1</i> | ✓ | | | | | | | | | ✓ | | | | | | ✓ | | | | | | | | ✓ | (Franco and Thauvin-Robinet, 2016) | |
| <i>PCDH15</i> | | | | | | | | | | | | | | | | | | ✓ | | | | | | ✓ | (Whatley et al., 2020) | |
| <i>PDE6D</i> | | | | | | | | | | ✓ | | | | | | | | | | | | | | | (Bachmann-Gagescu et al., 2020) | |
| <i>PDZD7</i> | | | | | | | | | | | | | | | | | | ✓ | | | | | | | (Booth et al., 2015) | |
| <i>PLK4</i> | | | | | | | | | | | ✓ | | | | | | | | | | | | | | (Martin et al., 2014) | |
| <i>POC1B</i> | | ✓ | | | | | | | | ✓ | | | | | | | | | | | | | | | (Beck et al., 2014) | |
| <i>RAB28</i> | | ✓ | | | | | | | | | | | | | | | | | | | | | | | (Gill et al., 2019) | |
| <i>RP1</i> | ✓ | | | ✓ | | | | | | | | | | | | | | | | | | | | | (Riera et al., 2020) | |
| <i>RP1L1</i> | ✓ | | | ✓ | | | | | | | | | | | | | | | | | | | | | (Noel and MacDonald, 2020) | |
| <i>RP2</i> | ✓ | | | | | | | | | | | | | | | | | | | | | | | | (Zhang et al., 2015) | |
| <i>RPGR</i> | ✓ | ✓ | | ✓ | | | | ✓ | | | | | | | | | | | | | | | | | (Shu et al., 2007) | |
| <i>RPGRIP1</i> | ✓ | ✓ | ✓ | | | | | | | | | | | | | | | | | | | | | | (Raghupathy et al., 2017) | |
| <i>RPGRIP1L</i> | ✓ | | ✓ | | | ✓ | | | | ✓ | | ✓ | | | | | | | ✓ | | | ✓ | | | (Wiegering et al., 2018) | |
| <i>SCAPER</i> | ✓ | | | | | | | | | | | | | | | | | | | | | | | | (Jauregui et al., 2019) | |
| <i>SDCCAG8</i> | | | | | | ✓ | | | | | | | | | | ✓ | | | | | | | | | (Suspitsin and Imyanitov, 2016) | |
| <i>SPATA7</i> | ✓ | | ✓ | | | | | | | | | | | | | | | | | | | | | | (Mackay et al., 2011) | |

| | Non-syndromic retinal ciliopathies | | | | Syndromic ciliopathies with retinal degeneration | | | | | | | | | | | | | | | Other ciliopathies without retinal degeneration | | | | | References | |
|----------------|------------------------------------|------|-----|----|--|-----|-----|------|-------|------|-------|-----|------|------|------|-----|------|-----|-------|---|------|------|-----|------|------------------------------------|------------------------|
| | RP | CORD | LCA | MD | AS | BBS | CED | CILD | CRDHL | JBTS | MCCRP | MKS | MORM | MRC5 | OFDS | SLS | SRTD | USH | COACH | LGMD | MKKS | NPHP | NSD | SGBS | | |
| <i>TBC1D32</i> | | | | | | | | | | | | | | | | | | | | | | | | | (Franco and Thauvin-Robinet, 2016) | |
| <i>TMEM107</i> | | | | | | | | | | ✓ | | ✓ | | | | | | | | | | | | | (Lambacher et al., 2016) | |
| <i>TMEM138</i> | | | | | | | | | | ✓ | | | | | | | | | | | | | | | (Bachmann-Gagescu et al., 2020) | |
| <i>TMEM216</i> | | | | | | | | | | ✓ | | ✓ | | | | | | | | | | | | | (Valente et al., 2010) | |
| <i>TMEM218</i> | | | | | | | | | | ✓ | | ✓ | | | | | | | | | | | | | (Van De Weghe et al., 2021) | |
| <i>TMEM231</i> | | | | | | | | | | ✓ | | ✓ | | | | | | ✓ | | | | | | | (Roberson et al., 2015) | |
| <i>TMEM237</i> | | | | | | | | | | ✓ | | | | | | | | | | | | | | | (Bachmann-Gagescu et al., 2020) | |
| <i>TMEM67</i> | | | | | | ✓ | | | | ✓ | | ✓ | | | | | | ✓ | | | | ✓ | | | (Radhakrishnan et al., 2019) | |
| <i>TOPORS</i> | ✓ | | | | | | | | | | | | | | | | | | | | | | | | (Chakarova et al., 2007) | |
| <i>TRIM32</i> | | | | | | ✓ | | | | | | | | | | | | | | ✓ | | | | | (Suspitsin and Imyanitov, 2016) | |
| <i>TTLL5</i> | | ✓ | | | | | | | | | | | | | | | | | | | | | | | (Gill et al., 2019) | |
| <i>TUBB4B</i> | | | ✓ | | | | | | | | | | | | | | | | | | | | | | (Luscan et al., 2017) | |
| <i>TUBGCP4</i> | | | | | | | | | | | ✓ | | | | | | | | | | | | | | (Scheidecker et al., 2015) | |
| <i>TULP1</i> | ✓ | | ✓ | | | | | | | | | | | | | | | | | | | | | | (Jacobson et al., 2014) | |
| <i>UNC119</i> | | ✓ | | | | | | | | ✓ | | | | | | | | | | | | | | | (Kobayashi et al., 2000) | |
| <i>USH1C</i> | | | | | | | | | | | | | | | | | | | | | | | | ✓ | (Whatley et al., 2020) | |
| <i>USH1G</i> | | | | | | | | | | | | | | | | | | | | | | | | ✓ | (Whatley et al., 2020) | |
| <i>USH2A</i> | ✓ | | | | | | | | | | | | | | | | | | | | | | | | (Whatley et al., 2020) | |
| <i>USH2C</i> | | | | | | | | | | | | | | | | | | | | | | | | | ✓ | (Whatley et al., 2020) |
| <i>USH2D</i> | | | | | | | | | | | | | | | | | | | | | | | | ✓ | (Whatley et al., 2020) | |

| | Non-syndromic retinal ciliopathies | | | | Syndromic ciliopathies with retinal degeneration | | | | | | | | | | | | | | Other ciliopathies without retinal degeneration | | | | | References | |
|--------------|------------------------------------|------|-----|----|--|-----|-----|------|-------|------|-------|-----|------|------|------|-----|------|-----|---|------|------|------|-----|------------|---------------------------------|
| | RP | CORD | LCA | MD | AS | BBS | CED | CILD | CRDHL | JBTS | MCCRP | MKS | MORM | MRCs | OFDS | SLS | SRTD | USH | COACH | LGMD | MKKS | NPHP | NSD | | SGBS |
| <i>WDPCP</i> | | | | | | ✓ | | | | | | | | | | | | | | | | | | | (Suspitsin and Imyanitov, 2016) |

^aFrom RetNet, OMIM, Syscilia, CiliaCarta and HGMD databases.

^bAS, Alström syndrome; BBS, Bardet-Biedl syndrome; CED, cranioectodermal dysplasia; CILD, ciliary dyskinesia; COACH, cerebellar vermis, hypo/aplasia, oligophrenia, congenital ataxia, ocular coloboma, and hepatic fibrosis; CORD, cone-rod dystrophy; CRDHL, cone-rod dystrophy and hearing loss; JBTS, Joubert syndrome; LCA, Leber congenital amaurosis; LGMD, limb-girdle muscular dystrophy; MCCRP, microcephaly and chorioretinopathy; MD, macular dystrophy; MKKS, McKusick-Kaufman syndrome; MKS, Meckel-Gruber syndrome; MORM, mental retardation, truncal obesity, retinal dystrophy, and micropenis; MRCs, microcornea-rod-cone dystrophy-cataract-posterior staphyloma syndrome; NPHP, nephronophthisis; NSD, non-syndromic deafness; OFDS, orofacialdigital syndrome; RP, retinitis pigmentosa; SGBS, Simpson-Golabi-Behmel syndrome; SLS, Senior-Løken syndrome; SRTD, short rib thoracic dysplasia with or without polydactyly; USH, Usher syndrome.

^cGenes are listed in alphabetic order: non-syndromic retinal ciliopathies causative genes are in blue, genes whose mutations result in syndromic retinal ciliopathies are in orange, and genes which, depending on the mutation, can cause both non-syndromic and syndromic retinal ciliopathies are in dark red. Non-syndromic retinal ciliopathies are colored in light blue, syndromic ciliopathies with retinal degeneration are in light orange, and other ciliopathies that do not show retinal degeneration as a phenotype are in light purple.