Supplementary Table 1: Occurrence and frequency of mutations in autosomal dominant (AD) cone and cone-rod dystrophy

STUDY	*AIPL1*	*CRX*	*GUCA1A*	*GUCY2D*	*PITPNM3*	*PROM1*	*PRPH2*	*RAX2*	*RIMS1*	*UNC119*
(Sohocki et al., 2000b)	1/15									
(Boulanger-scemama et al., 2015)	2/96	4/96	1/96	5/96		9/96	2/96			
(Freund et al., 1997)		2/30								
(Swain et al., 1997)		4								
(Sohocki et al., 1998)		3/30								
(Jacobson et al., 1998)		1								
(Sankila et al., 2000)		1								
(Sohocki et al., 2000a)		4/67								
(Rivolta et al., 2001)		0/88								
(Lines et al., 2002)		1								
(Itabashi et al., 2004)		1								
(Henderson et al., 2007)		1/29								
(Paunescu et al., 2007)		1								
(Kitiratschky et al., 2008b)		1								
(Kitiratschky et al., 2011)		1/7	3/22	3/7			2/22			
(Kohl et al., 2012)		2/52	4/52	12/52		1/52	6/52			
(Huang et al., 2012)		3/130								
(Lu et al., 2015)		1								
(Huang et al., 2016)		3/163	2/163	5/163						1/163
(Oishi et al., 2016)		2/43		1/43		2/43	1/43			
(Birtel et al., 2018)		4/251	3/251	6/251		6/251	29/251			
(Payne et al., 1998)			1							
(Downes et al., 2001a)			3							
(Wilkie et al., 2001)			1							
(Nishiguchi et al., 2004)			1/119							
(Jiang et al., 2005)			1							
(Michaelides et al., 2005)			1							
(Sokal et al., 2005)			1							
(Jiang et al., 2008)			1							
(Kitiratschky et al., 2009)			4/24							
(Kamenarova et al., 2013)			2							
(Huang et al., 2013a)			1							
(Nong et al., 2014)			1							
(Manes et al., 2017)			2							
(Marino et al., 2018)			1							
(Kelsell et al., 1998)				4						
(Gregory-evans et al., 2000)				1						
(Weigell-weber et al., 2000)				1						

STUDY	*AIPL1*	*CRX*	*GUCA1A*	*GUCY2D*	*PITPNM3*	*PROM1*	*PRPH2*	*RAX2*	*RIMS1*	*UNC119*
(Van ghelue et al., 2001)				1						
(Downes et al., 2001b)				4						
(Payne et al., 2001)				3/40						
(Udar et al., 2003)				2						
(Ito et al., 2004a)				3/38						
(Ito et al., 2004b)				2						
(Yoshida et al., 2006)				1						
(Smith et al., 2006)				1						
(Kitiratschky et al., 2008c)				11/27						
(Small et al., 2008)				1						
(Garcia-hoyos et al., 2011)				2/22						
(Xiao et al., 2011)				1						
(Xu et al., 2013)				1						
(Huang et al., 2013b)				2/130			1/130			1/130
(Zhao et al., 2013)				1						
(Alapati et al., 2014)				6/54						
(Lazar et al., 2015)				2/6						
(Jiang et al., 2015)				7/15						
(Köhn et al., 2007)					2					
(Köhn et al., 2010)					5/163					
(Yang et al., 2008)						3				
(Michaelides et al., 2010)						5				
(Eidinger et al., 2015)						1				
(Mayer et al., 2016)						1				
(Wawrocka et al., 2018)						1/18	1/18			
(Nakazawa et al., 1996a)							4			
(Nakazawa et al., 1996b)							1			
(Renner et al., 2009)							3			
(Yang et al., 2015)								1		
(Johnson et al., 2003)									1	
(Kobayashi et al., 2000)										1/20
OVERALL MUTATION FREQUENCY	3/111 (1.2%)	40/997 (15.6%)	34/743 (13.2%)	89/965 (34.6%)	7/165 (2.7%)	29/470 (11.3%)	50/620 (19.5%)	1/1 (0.4%)	1/1 (0.4%)	3/313 (1.2%)

Note: Denominators represent the total number of unrelated probands with a clinical diagnosis of progressive cone dystrophy (COD) or cone-rod dystrophy (CORD) in each patient cohort. Numerators represent the number of patients with disease secondary to pathogenic variant/s in each respective gene. Non-fractional integers represent the number of unique pathogenic variants identified in a gene from pedigree analysis.

Percentages of overall mutation frequency represent the proportion of cases each gene is responsible for from the total number of identified genetic variants in a single mode of inheritance.

Supplementary Table 2: Occurrence and frequency of mutations in autosomal recessive (AR) cone and cone-rod dystrophy

STUDY	*ABCA4*	*ADAM9*	*C21ORF2*	*C8ORF37*	*CACNA2D4*	*CDHR1*	*CEP78*	*CERKL*	*CNGA3*	*CNGB3*	*KCNV2*	*PDE6C*	*PDE6H*	*POC1B*	*RAB28*	*RPGRIP1*	*SEMA4A*	*TTLL5*
(Cremers et al., 1998)	1																	
(Maugeri et al., 2000)	13/20																	
(Papaioannou et al., 2000)	4/8																	
(Birch et al., 2001)	11/37																	
(Briggs et al., 2001)	5/8																	
(Paloma et al., 2001)	2																	
(Ducroq et al., 2002)	13/55																	
(Paloma et al., 2002)	1																	
(Fishman et al., 2003)	16/30																	
(Klevering et al., 2004a)	1																	
(Klevering et al., 2004b)	18/54																	
(Simonelli et al., 2004)	1																	
(Klevering et al., 2005)	2																	
(Ducroq et al., 2006)	1																	
(Stenirri et al., 2006)	1																	
(Singh et al., 2006)	2/14																	
(Michaelides et al., 2007)	4																	
(Yzer et al., 2007)	1																	
(Kitiratschky et al., 2008a)	20/64																	
(Ernest et al., 2009)	3																	
(Xi et al., 2009)	1																	
(Aguirre-lamban et al., 2009)	1/5																	
(Littink et al., 2010)	33/123							2/123			1/123							
(Thiadens et al., 2012)	25/155									3/90	4/90							
(Huang et al., 2013c)	1/47									3/47		2/47				1/47		
(Bocquet et al., 2013)	16/34																	
(Riveiro-alvarez et al., 2013)	26/86																	
(Alapati et al., 2014)	15/54																	
(Boulanger-scemama et al., 2015)	23/96		2/96			1/96		1/96				4/96				1/96	2/96	
(Huang et al., 2016)	6/163		1/163						53/163	3/163	1/163	3/163				2/163		
(Oishi et al., 2016)	4/43					1/43												
(Birtel et al., 2018)	94/251					6/251		2/251						1/251			1/251	
(Parry et al., 2009)		4																
(El-haig et al., 2014)		1																
(Hull et al., 2014)		1																

STUDY	*ABCA4*	*ADAM9*	*C21ORF2*	*C8ORF37*	*CACNA2D4*	*CDHR1*	*CEP78*	*CERKL*	*CNGA3*	*CNGB3*	*KCNV2*	*PDE6C*	*PDE6H*	*POC1B*	*RAB28*	*RPGRIP1*	*SEMA4A*	*TTLL5*
(Khan et al., 2015)			3															
(Suga et al., 2016)			2															
(Estrada-cuzcano et al., 2012)				4														
(Lazar et al., 2015)				1/6		1/6												
(Rahner et al., 2016)				1														
(Katagiri et al., 2016)				2														
(Wycisk et al., 2006)					1/34													
(Ba-abbad et al., 2016)					2													
(Ostergaard et al., 2010)						1												
(Henderson et al., 2010)						2												
(Cohen et al., 2012)						1												
(Fu et al., 2018)						1												
(Nikopoulos et al., 2016)							2											
(Namburi et al., 2016)							5											
(Fu et al., 2017)							2											
(Auslender et al., 2007)								7/21										
(Ali et al., 2008)								1										
(Aleman et al., 2009)								3										
(Wissinger et al., 2001)									3/48									
(Shaikh et al., 2015)									1									
(Michaelides et al., 2004)										4								
(Thiadens et al., 2010)										3/60								
(Wu et al., 2006)											10/11							
(Thiagalingam et al., 2007)											8							
(Salah et al., 2008)											3							
(Wissinger et al., 2008)											11							
(Wissinger et al., 2011)											4/154							
(Fujinami et al., 2013)											2							
(Thiadens et al., 2009)												1/104						
(Piri et al., 2005)													1					
(Durlu et al., 2014)														1				
(Roosing et al., 2014)														2				
(Jin et al., 2018)														1				
(Roosing et al., 2013)															2/617			
(Riveiro-álvarez et al., 2015)															2/109			
(Hameed et al., 2003)																2		
(Abid et al., 2006)																	2/25	
(Sergouniotis et al., 2014)																		4/83

STUDY	*ABCA4*	*ADAM9*	*C21ORF2*	*C8ORF37*	*CACNA2D4*	*CDHR1*	*CEP78*	*CERKL*	*CNGA3*	*CNGB3*	*KCNV2*	*PDE6C*	*PDE6H*	*POC1B*	*RAB28*	*RPGRIP1*	*SEMA4A*	*TTLL5*
(Bedoni et al., 2016)																		6
OVERALL MUTATION FREQUENCY	365/ 1366 (62.2%)	6/6 (1.0%)	8/264 (1.4%)	8/13 (1.4%)	3/36 (0.5%)	14/401 (2.4%)	9/9 (1.5%)	16/495 (2.7%)	57/212 (9.7%)	16/364 (2.7%)	44/565 (7.5%)	10/410 (1.7%)	1/1 (0.2%)	5/255 (0.9%)	4/726 (0.7%)	6/308 (1.0%)	5/372 (0.9%)	10/89 (1.7%)

Supplementary Table 3: Occurrence and frequency of mutations in X-linked (XL) cone and cone-rod dystrophy

STUDY	*CACNA1F*	*OPN1LW / OPN1MW*	*RPGR*
(Jalkanen et al., 2006)	1		
(Huang et al., 2013c)	1/47		2/47
(Hauke et al., 2013)	1		
(Huang et al., 2016)	4/163		
(Gardner et al., 2010)		2	
(Gardner et al., 2012)		1	
(Demirci et al., 2002)			3
(Yang et al., 2002)			2
(Ebenezer et al., 2005)			2/6
(Ruddle et al., 2009)			3
(Thiadens et al., 2012)			7/8
(Bocquet et al., 2013)			2/10
(Birtel et al., 2018)			1/251
(Wawrocka et al., 2018)			5/18
OVERALL MUTATION FREQUENCY	7/212 (18.9%)	3/3 (8.1%)	27/348 (73.0%)

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