Retinal ciliopathies [1] can occur as isolated retinal conditions or in combination with other ciliopathies which encompass ectodermal, cerebrorenal and metabolic disorders and are caused by a wide array of genetic mutations. [1], [2], [3], [4], [5], [6], [7], [8], [9], [10], [11], [12], [13], [14], [15], [16], [17], [18], [19]

Cone-rod ciliopathies: Listed by IK Jalili, 2010. Revised November 2011. http://www.jalili.co/CRC/CRC_T2011.pdf

A) Ectodermal Disorders

Amelogenesis imperfecta
Jalili syndrome (OMIM 217080) [20], [21]
CNNM4: 2q11 [5], [6]

Retinal phenotype A

Gaza A	c.599→A; p.Ser200yr [5]
Kosovar 1	c.1312dupC; p.Leu438ProfsX9 [5], [22]
Kosovar 2	c.1312dupC; p.Leu438ProfsX9
Kosovar 3	c.1312dupC; p.Leu438ProfsX9 [23]
Lebanese	c.707G→A; p.R236Q [6]
Unknown	c.971T→C; p.L324P [6]

Retinal Phenotype B

Gaza B [21]	c.1813C→T; p.Arg605X [5]
Scottish	c.971T→C; p.Leu324Pro / c.1690C→T; p.Gln564X [5]
Turkish	c.586T→C; p.Ser196Pro [5]

Retinal phenotype unspecified

Iranian	c.1-?_1403+?del [5]
Guatemalan	c.2149C→T; p.Gln717X) / c.62_145del; pLeu21HisfsX185 [5]

Hypertrichosis with trichomegaly (OMIM 204110)
Cone-rod congenital amaurosis [1]

Hypotrichosis (OMIM 601553)
Hereditary Juvenile macular degeneration (HJMD)
CDH3 (16q22.1) [7]
Group1: Single nucleotide deletion (981delG) encoding P-cadherin [8]
Group 2: Missense mutation (R503H) [9]
Alopecia of scalp and eyelashes [10]
Aplasia cutis congenital with high myopia (OMIM 600360) [11]

B) Dysmorphic Syndromes

Spondylometaphyseal dysplasia [12], [13]
cleft lip [14]

C) Metabolic Dysfunctions

Thiamine-responsive megaloblastic anaemia (TRMA, Rogers syndrome) [15] (OMIM 249270)
SLC19A2 loc 1q23.3

D) Cerebro-Renal Disorders

Bardet Biedl syndrome and Variants (OMIM 209900) [16], [24] [25]

BBS1 (BBS2L2): 11q13.1
BBS2 (BBS, MGC20703): 16q21 (c.565C>T; p.ArgR189Stop)
ARLS6 (*BSS3*): 3q11.2
BBS4: 15q22.3-q23
BBS5: 2q31.1 (c.123delA; p.Gly42GlufsX11)
MKKS (BBS6): 20p12
BBS7 (BBS2L1): 4q27 (g.47247455_47267458del20004insATA; p.Met284LysfsX7)
TTC8 (BSS8): 14q31.3 (1- c.459+1G>A; p.Pro101LeufsX12); (c.355_356insGGTGGAAGGCCAGGCA; p.Thr124ArgfsX43)
BBS9 (B1, C18, D1, MGC118917): 7p14
BBS10: 10 D1 (C91LfsX4(C91fsX95)
TRIM32 (BBS11): 9q33.1
BSS12 (C4orf24): 4q27
MKS1 (BBS13): 17q22
CEP290 (BBS14): 12q21.32
TMEM67 (HBTS6, MKS3): 8q22.1

Biemond syndrome (plus iris coloboma)

Alström syndrome (OMIM 203800) [17]
NPHP1 ; AHI1 ; CEP290 (NPHP6) ; TMEM67 (MKS); JBTS1; CORS2 (JBTS2);

Spinocerebellar ataxia type 7 (OMIM 164500) [18], [19]; *ATXN7 (ADCAII, OPCA3, SCA7):* 3p21.1-p12

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