

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; p.Leu21HisfsX185⁽⁵⁾

Hypertrichosis with trichomegaly (OMIM 204110)

Cone-rod congenital amaurosis⁽¹⁾

Hypotrichosis (OMIM 601553)

Hereditary Juvenile macular degeneration (HJMD)
CDH3 (16q22.1)⁽⁷⁾
Group 1: 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 (BBS3): 3q11.2
BBS4: 15q22.3-q23
BBS5: 2q31.1 (c.123delA; p.Gly42Glu fsX11)
MKKS (BBS6): 20p12
BBS7 (BBS2L1): 4q27
 (g.47247455_47267458del20004insATA; p.Met284Lys fsX7)
TTC8 (BBS8): 14q31.3 (1- c.459+1G>A; p.Pro101Leu fsX12);
 (c.355_356insGGTGGGAAGGCCAGGCA; p.Thr124Arg fsX43)
BBS9 (B1, C18, D1, MGC118917): 7p14
BBS10: 10 D1 (C91LfsX4(C91fsX95)
TRIM32 (BBS11): 9q33.1
BBS12 (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 ; *AH1* ; *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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