

36 Diseases, 54 Genes

(self test)

Big 12 (14 genes)

I - Photoreceptor Diseases (6)

Nonsyndromic

Acquired and/or Progressive

XL

AR

AD

Retinitis Pigmentosa

Cone or Cone-rod Dystrophy

Congenital and/or Stationary

LCA Spectrum

Achromatopsia

Syndromic

Usher Syndrome

Bardet Biedl Syndrome

II - Macular Dystrophies (3)

AR Stargardt Disease

Best Disease

Pattern Dystrophy

III - Third Branch (3)

Choroidopathies

Choroideremia

Retinoschisis

XL Retinoschisis

Optic Neuropathies

Nonsyndromic

Dominant Optic Atrophy

Big 14 (2 more genes)

SECORD

ECORD

Next 8 genes:

ARRP (FEMP)

ADRP (PRRP)

XLRP

Usher

Next 22 diseases and 23 genes (**bold** indicates diseases in the Big 14 on the previous page)

Macular Dystrophies: **Singapore beaches provide** pleasant memories on summer nights sipping mojitos.

Syndromic Photoreceptor Diseases: **yoU better** be sure Jan makes all her children attend (Stone Rounds).

PXE	_____	Batten	_____
MIDD	_____	Senior Loken	_____
OMD	_____	Joubert	_____
Stargardt (AD)	_____..._____	MCL	_____
NCMD	_____	Ataxia	_____
Sorsby	_____	Heimler	_____
Malattia Leventinese	_____	Cohen	_____
		Anemia	_____

Third Branch Diseases: **CRO** TV AF

Congenital Stationary Photoreceptor Diseases: **LSEA** BCC

Tumors	_____	BCM	_____
Vitreoretinopathies	_____	CSNB	_____
Albinism	_____	CSSD	_____
Foveal Hypoplasia	_____		

Last 7 genes (CRINGe) (Mercer)

LCA _____

CRD _____

36 Diseases, 54 Genes

(answers)

Big 12 (14 genes)

I - Photoreceptor Diseases (6)

Nonsyndromic

Acquired and/or Progressive	XL	AR	AD
Retinitis Pigmentosa	<u>RPGR</u>	<u>USH2A</u>	<u>RHO</u>
Cone or Cone-rod Dystrophy	<u>RPGR</u>	<u>ABCA4</u>	<u>GUCA1A</u>
Congenital and/or Stationary			
LCA Spectrum	<u>CEP290</u>		
Achromatopsia	<u>CNGB3</u>	<u>CNGA3</u>	
Usher Syndrome	<u>USH2A</u>		
Bardet Biedl Syndrome	<u>BBS1</u>		

II - Macular Dystrophies (3)

AR Stargardt Disease	<u>ABCA4</u>
Best Disease	<u>BEST1</u>
Pattern Dystrophy	<u>PRPH2</u>

III - Third Branch (3)

Choroidopathies	
Choroideremia	<u>CHM</u>
Retinoschisis	
XL Retinoschisis	<u>RS1</u>
Optic Neuropathies	
Nonsyndromic	
Dominant Optic Atrophy	<u>OPA1</u>

Big 14 (2 more genes)	SECORD	<u>RDH12</u>
	ECORD	<u>CRB1</u>

Next 8 genes:	ARRP (FEMP)	<u>FAM161a</u>	<u>EYS</u>	<u>MAK</u>	<u>PDE6B</u>
	ADRP (PRRP)	<u>PRPF31</u>	<u>RP1</u>		
	XLRP	<u>RP2</u>			
	Usher	<u>MYO7A</u>			

Next 22 diseases and 23 genes (**bold** indicates diseases in the Big 14 on the previous page)

Macular Dystrophies: **Singapore beaches provide** pleasant memories on summer nights sipping mojitos.

Syndromic Photoreceptor Diseases: **yoU better** be sure Jan makes all her children attend (Stone Rounds).

PXE	<u>ABCC6</u>	Batten	<u>CLN3</u>
MIDD	<u>mt-TL1</u> (3243)	Senior Loken	<u>IQCB1</u> <u>NPHP1</u>
OMD	<u>RP1L1</u>	Joubert	<u>INPP5E</u>
Stargardt (AD)	<u>ELOVL4</u> <u>PROM1</u>	MCL	<u>KIF11</u>
NCMD	<u>PRDM13</u>	Ataxia	<u>FLVCR1</u>
Sorsby	<u>TIMP3</u>	Heimler	<u>PEX1</u>
Malattia Leventinese	<u>EFEMP1</u>	Cohen	<u>VPS13B</u>
		Anemia	<u>TRNT1</u>

Third Branch Diseases: **CRO** TV AF

Congenital Stationary Photoreceptor Diseases: **LSEA** BCC

Tumors	<u>VHL</u>	Blue Cone Monochromacy	<u>OPN1LW/MW</u>
Vitreoretinopathies	<u>COL2A1</u>	Congenital Stationary Night Blindness	<u>NR2E3</u>
Albinism	<u>TYR</u>	Congenital Stationary Synaptic Dysfunction	<u>CACNA1F</u>
Foveal Hypoplasia	<u>PAX6</u>		

Last genes (CRNGe) (Mercer)

LCA CRX RPE65 NMNAT1 GUCY2D

CRD MERTK CERKL