### 36 Diseases, 54 Genes

(self test)

Big 12 (14 genes)

I - Photoreceptor Diseases (6)

**Nonsyndromic**

<table>
<thead>
<tr>
<th>Acquired and/or Progressive</th>
<th>XL</th>
<th>AR</th>
<th>AD</th>
</tr>
</thead>
<tbody>
<tr>
<td>Retinitis Pigmentosa</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Cone or Cone-rod Dystrophy</td>
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</tbody>
</table>

**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 9 genes:

| ARRP (FEMP)                                  |    |    |    |
| ADRP (PR<sub>2</sub>RP)                      |    |    |    |
| XLRP                                         |    |    |    |
| Usher                                        |    |    |    |
Next 22 diseases and 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

Retinitis Pigmentosa
Retinitis Pigmentosa
Retinitis Pigmentosa

Cone or Cone-rod Dystrophy
Cone or Cone-rod Dystrophy
Cone or Cone-rod Dystrophy

Congenital and/or Stationary

LCA Spectrum
LCA Spectrum
LCA Spectrum

Achromatopsia
Achromatopsia
Achromatopsia

Usher Syndrome
Usher Syndrome
Usher Syndrome

Bardet Biedl Syndrome
Bardet Biedl Syndrome
Bardet Biedl Syndrome

II - Macular Dystrophies (3)

AR Stargardt Disease
AR Stargardt Disease
AR Stargardt Disease

Best Disease
Best Disease
Best Disease

Pattern Dystrophy
Pattern Dystrophy
Pattern Dystrophy

III - Third Branch (3)

Choroidopathies
Choroidopathies
Choroidopathies

Choroideremia
Choroideremia
Choroideremia

Retinoschisis
Retinoschisis
Retinoschisis

XL Retinoschisis
XL Retinoschisis
XL Retinoschisis

Optic Neuropathies
Optic Neuropathies
Optic Neuropathies

Nonsyndromic
Nonsyndromic
Nonsyndromic

Dominant Optic Atrophy
Dominant Optic Atrophy
Dominant Optic Atrophy

Big 14 (2 more genes)

SECORD
SECORD
SECORD

ECORD
ECORD
ECORD

RDH12
RDH12
RDH12

CRB1
CRB1
CRB1

Next 9 genes:

ARRP (FEMP) ARRP (FEMP) ARRP (FEMP)

FAM161a
FAM161a
FAM161a

EYS
EYS
EYS

MAK
MAK
MAK

PDE6B
PDE6B
PDE6B

PRPF31
PRPF31
PRPF31

PROM1
PROM1
PROM1

RP1
RP1
RP1

XLRP
XLRP
XLRP

RP2
RP2
RP2

Usher
Usher
Usher

MYO7A
MYO7A
MYO7A
Next 22 diseases and 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).

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<th>PEXP1</th>
<th>ABCC6</th>
<th>Batten</th>
<th>CLN3</th>
</tr>
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<tbody>
<tr>
<td>MIDD</td>
<td>mt-TL1 (3243)</td>
<td>Senior Loken</td>
<td>NPHP1</td>
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<tr>
<td>OMD</td>
<td>RP1L1</td>
<td>Joubert</td>
<td>INPP5E</td>
</tr>
<tr>
<td>Stargardt (AD)</td>
<td>ELOVL4</td>
<td>MCL</td>
<td>KIF11</td>
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<tr>
<td>NCMD</td>
<td>PRDM13</td>
<td>Ataxia</td>
<td>FLVCR1</td>
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<tr>
<td>Sorsby</td>
<td>TIMP3</td>
<td>Heimler</td>
<td>PEX1</td>
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<tr>
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<td>EFEMP1</td>
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| Third Branch Diseases: **CRO TV AF**
Congenital Stationary Photoreceptor Diseases: **LSEA BCC** |

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<th>Tumors</th>
<th>VHL</th>
<th>Blue Cone Monochromacy</th>
<th>OPN1LW/MW</th>
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<tr>
<td>Vitreoretinopathies</td>
<td>COL2A1</td>
<td>Congenital Stationary Night Blindness</td>
<td>NR2E3</td>
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<tr>
<td>Albinism</td>
<td>TYR</td>
<td>Congenital Stationary Synaptic Dysfunction</td>
<td>CACNA1F</td>
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Last 7 genes (CRINGe) (Mercer)

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<th>RPE65</th>
<th>IQCB1</th>
<th>NMNAT1</th>
<th>GUCY2D</th>
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<td>MERTK</td>
<td>CERKL</td>
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