



## **Syndromic Features to Consider**

Deafness, polydactyly, obesity, diabetes, abnormal cognition, developmental delay, seizures, ataxia, dysarthria, other brain abnormalities, hypertension, heart disease, malformation and / or arrhythmias, nocturia, other kidney abnormalities, abnormal dentition, cleft palate, dysmorphic facies, abnormal bruising or bleeding, frequent infections, malabsorption, muscle weakness.

**[www.carverlab.org](http://www.carverlab.org)**

## Big 14 (87%)

### I - Photoreceptor Disease

#### A - Isolated

##### 1 - Acquired/Progressive

a - Retinitis Pigmentosa

b - Cone & C/R Dystrophy

##### 2 - Congenital/Stationary

a - LCA

b - SECORD

c - ECORD

d - Achromatopsia

#### B - Syndromic

##### 1 - Usher Syndrome

##### 2 - Bardet-Biedl Syndrome

### II - Macular Diseases

#### A - AR Stargardt Disease

#### B - Best Disease

#### C - Pattern Dystrophy

### III - Third Branch Disorders

#### A1 - Choroideremia

#### B1 - XL Retinoschisis

#### C1a - Dominant Optic Atrophy