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Diagnosis & Co-morbidities

Date of Diagnosis


Retinal Diagnosis
- Achromatopsia
- Age-Related Macular Degeneration - Dry
- Age-Related Macular Degeneration - Wet
- Age-Related Macular Degeneration - Both
- Age-Related Macular Degeneration - Unspecified
- Best Disease
- Biette crystalline Corneoretinal Dystrophy
- Blue Cone Monochromacy
- Choroideremia
- Cone Dystrophy
- Cone-Rod Dystrophy
- Congenital Stationary Night Blindness
- Gyrate Atrophy
- Juvenile Inherited Macular Degeneration
- Leber Congenital Amaurosis
- Mallatia Leventinese
- North Carolina Macular Dystrophy
- Oguchi Disease
- Pattern Dystrophy
- Retinitis Pigmentosa
- Retinitis Pigmentosa - Atypical
- Retinitis Punctata Albescens
- Retinoschisis
- Rod-Cone Dystrophy
- Rod Monochromatism
- Sorsby Fundus Dystrophy
- Stargardt Disease
- Unknown

Syndromic Diagnosis:
- Alstrom Syndrome
- Bardet-Biedl Syndrome (Laurence-Moon)
- Bassen-Kornzweig Syndrome
- Batten Disease
- Enhanced S-Cone Syndrome
- Joubert Syndrome
- Refsum Syndrome
- Senior-Loken Syndrome
- Usher Syndrome
- Unknown
- Other
- Not Applicable
Associated Medical Conditions
Not Available
No Significant Medical Nonocular Issue
Hearing Deficit
High Blood Pressure
Neurological Disease
Kidney Anomaly
Diabetes
Developmental Delay
Cognitive Impairment
Digit Anomaly
Psychiatric Disease
Asthma
Psoriasis
Crohn's Disease
Multiple Sclerosis
Rheumatoid Arthritis

Secondary Retinal Diagnosis
Achromatopsia
Age-Related Macular Degeneration - Dry
Age-Related Macular Degeneration - Wet
Age-Related Macular Degeneration - Both
Age-Related Macular Degeneration - Unspecified
Best Disease
Biette crystalline Corneoretinal Dystrophy
Blue Cone Monochromacy
Choroideremia
Cone Dystrophy
Cone-Rod Dystrophy
Congenital Stationary Night Blindness
Gyrate Atrophy
Juvenile Inherited Macular Degeneration
Leber Congenital Amaurosis
Mallatia Leventinese
North Carolina Macular Dystrophy
Oguchi Disease
Pattern Dystrophy
Retinitis Pigmentosa
Retinitis Pigmentosa - Atypical
Retinitis Punctata Albescens
Retinoschisis
Rod-Cone Dystrophy
Rod Monochromatism
Sorsby Fundus Dystrophy
Stargardt Disease
Unknown
Not Applicable
ERG (Full Field)

Date of Most Recent Examination

Summary of Results
    Normal
    Abnormal
    Not Recordable

ISCEV Standard (if reported)
    Yes
    No

Scotopic Rod b Wave - Amplitude
    OD
    OS
    Option for each:
        Unselected
        Normal
        Abnormal
        Not Recordable

Scotopic Rod b Wave - Implicit Time
    OD
    OS
    Option for each:
        Unselected
        Normal
        Abnormal
        Not Recordable

Rod/cone a-wave - Amplitude
    OD
    OS
    Option for each:
        Unselected
        Normal
        Abnormal
        Not Recordable

Rod/cone b-wave - Amplitude
    OD
    OS
    Option for each:
        Unselected
        Normal
        Abnormal
        Not Recordable
Cone b-wave - Amplitude
  OD
  OS
  Option for each:
    Unselected
    Normal
    Abnormal
    Not Recordable

Cone b-wave - Implicit Time
  OD
  OS
  Option for each:
    Unselected
    Normal
    Abnormal
    Not Recordable

Photopic 30 Hz Flicker - Amplitude
  OD
  OS
  Option for each:
    Unselected
    Normal
    Abnormal
    Not Recordable

Photopic 30 Hz Flicker - Implicit Time
  OD
  OS
  Option for each:
    Unselected
    Normal
    Abnormal
    Not Recordable

EZ Width

Date of Most Recent Measurement
Is the end of the EZ band beyond the edge of the scan?
  Yes
  No

Is there a visible EZ band?
  Detectable
  Non-detectable

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EZ band width (in mm)
  horizontal midline (mm)
  vertical midline (mm)

Genetic Diagnosis

Is there a family inheritance pattern?
  Yes
  No
  Unknown
  Waiting for Results

What is the suspected mode of inheritance?
  Unknown
  Autosomal Dominant (AD)
  Autosomal Recessive (AR)
  X-Linked Recessive (XL)
  Isolated/Simplex/Sporadic

Number of disease related mutations identified?

For Each Disease Related Mutation

Date of Molecular Diagnosis

Reference Gene Sequence
  Free text e.g. (example: NM_006445)

Gene Name

Zygosity
  homozygous (two identical mutations reported)
  heterozygous (one mutation reported)
  compound heterozygote (two different mutations reported)
  hemizygous (one X-linked mutation reported in a male)
  unknown/other

Disease Causing?
  Pathogenic/Deleterious
  Probable
  Unlikely
  Unknown
  Not

cDNA sequence change
  Free text e.g.  c.2299delG, c.581G>A, etc.

Protein sequence change
  Free text e.g.  p.(Glu767Serfs*21, P.(Trp194*), etc.
**Impact of mutation (if reported)**
Free text e.g. mis-splicing of exon 14, etc.

**Additional annotations not previously captured or comments**
Free text

**Have additional disease related mutations been reported for this patient?**
- Yes
- No

**Kinetic Visual Field**

**Date of Most Recent Examination**

**Instrument Used**
- Goldman
- Octopus 101
- Octopus 900

**Type**
- I4e
- II4e
- III4e
- IV4e
- V4e
- Not Done

**Result**
- OD
- OS

Option for each:
- Normal
- Central Scotoma
- Para Central Scotoma
- Annular Scotoma
- Central Island
- Peripheral Island

**Vertical diameter**
- OD
- OS

Option for each:
- Normal
- ≤ 5 degrees
- ≤ 10 degrees
- ≤ 20 degrees
- ≤ 50 degrees
- ≤ 100 degrees
- ≤ 120 degrees
Horizontal diameter

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<td>≤ 50 degrees</td>
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<td>≤ 100 degrees</td>
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<td>≤ 120 degrees</td>
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Light/Dark Adaptation

Date of Most Recent Examination
Result
Normal
Abnormal

mfERG

Date of Most Recent Examination
ISCEV Standard (if reported)
Yes
No

Result

<table>
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Ocular Assessment

Date of Most Recent Examination
Cataract

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Cataract Severity
OD
OS
Option for each:
  Mild
  Moderate
  Severe
  Not Applicable

Cornea
OD
OS
Option for each:
  Normal
  Crystals
  Keratoconus

Optic Disc
OD
OS
Option for each:
  Normal
  Pale
  Abnormal
  Optic Atrophy
  Drusen

Macula
OD
OS
Option for each:
  Normal
  Abnormal
  Not Done
  Atrophy
  Edema
  Schisis
  Bulls Eye
  Wrinkling
  Drusen
  Flecks
  Exudates
  Other Deposits
  RPE Changes

Retina - OD
OD
OS
Option for each:
Normal
Abnormal
Not Done
Retinoschisis
Bone Spiculing
Pigment Clumping
White Dots
Flecks
Drusen
Retinal detachment
Mottling

**IVFA**

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**Autofluorescence**

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**OCT**

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| Option for each: | Normal | Abnormal | Not Done
|                | Schisis| Edema   |
MD (Mean Defect)
- OD: Free text (decibels)
- OS: Free text (decibels)

Static Visual Field

Date of Most Recent Examination

Name of Instrument Used
- Humphrey
- Octopus 101
- Octopus 900

Threshold
- SITA
- 4-2-1
- GATE

Stimulus Size
- 3
- 4
- 5
- 6

Type
- 30-2
- 24-2
- 10-2
- Full Field
- Custom Field
- Not Done

Result
- OD
- OS

Option for each:
- Central Scotoma
- Normal
- Para Central Scotoma
- Annular Scotoma
- Concentric Constriction
- Central Island

MD (Mean Defect)
- OD: Free text (decibels)
- OS: Free text (decibels)
Visual Acuity

Date of Most Recent Examination

Correction

OD
OS

Option for each:
High Hyperopic (more than +4)
Mild Hyperopic (+2 to +4)
Emmetropic
Mild Myopia
Moderate Myopia (-2 to -5)
High Myopia (more than -6)
None

Astigmatism

OD
OS

Option for each:
None
1.5D
1.5 - 3.0D
>3.0D

Best Corrected Visual Acuity (BCVA)

Select the menu that represents the measurement distance (20ft, 10ft, 6m or 3m) used during testing. For very low vision, abbreviations used below are: CF count fingers, HM hand motion, LP light perception, NLP no light perception.

OD
OS

Option for each:

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