Dear Doctor,

Your patient has a rare syndrome, Bosch-Boonstra-Schaaf optic atrophy syndrome (BBSOAS), an autosomal dominant condition caused by a disruption in the NR2F1 gene.

Features present at birth:
- Hypotonia
- Oromotor dysfunction
- Vision problems such as nystagmus and poor tracking

Features which develop over time:
- Development delay/intellectual disability
- Seizures
- Autism Spectrum disorder
- Vision impairment – 90%
  - Due to optic nerve abnormalities +/- brain based vision impairment
  - Optic nerve atrophy or pallor - 82%
  - Optic nerve hypoplasia - 82%
    - Not the classic found in septo-optic dysplasia, we found slightly smaller than normal sized optic nerve heads, no severe hypoplasia
  - Cortical visual impairment - 68%
- Other ocular features:
  - A/hypolacrimalia (decreased amount of emotional and reflex tearing) - 78%
  - Manifest latent nystagmus (infantile onset of manifest nystagmus, +/- latent component, or latent nystagmus alone - 52%
    - Often improved over time
  - Significant refractive errors
  - Amblyopia
Suggested evaluation

Targeted History

- History of abnormal visual function?
  - Is it suggestive of CVI?
    - Abnormal vision with normal or mildly abnormal ocular findings
  - As infants, light gazing
  - Difficulty locating objects in a crowded field
  - Variable visual attention, especially in unfamiliar environment
  - Preference for high contrast objects

- History of nystagmus?
- History of abnormal volume of reflex tearing?

Targeted Exam

- Visual acuity (test which is appropriate for patient’s age and understanding)
  - Teller acuity cards, Allen pictures, LEA, HOTV, Snellen
  - Color vision (indicator of optic nerve function)

- External exam
  - Manifest nystagmus? Latent component?

- Pupil reactivity
  - Poorly reactive, APD?

- Visual fields
  - Nonspecific abnormalities due to optic nerve abnormalities or CVI

- Intraocular Pressure
  - Glaucoma was not a finding in our pts

- Anterior segment
  - Dry eye findings?
    - Not a feature in our pts

- Optic nerve
  - Atrophy? mild or severe?
  - Mild hypoplasia?
    - Normal optic nerve diameter
      - 3 disc diameters fit in between the center of the optic nerve and the fovea
    - Mild hypoplasia
      - >3 disc diameters fit in between the center of the optic nerve and the fovea
Suggested In-office testing, if able:

- Fundus photography to document optic nerve health
  - Document if pallor exists and the distance between center of the optic nerve border and fovea (rule out mild hypoplasia)
- OCT
  - RNFL to document health of the optic nerve
  - GCL as surrogate for VFs
- Visual fields
  - Confrontation, if able, automated perimetry
    - We were unable to perform VF testing on any patient

Thank you for helping our BBSOAS patients and families, Jane Edmond, MD

For more information, visit NR2F1 Foundation: nr2f1.org