

Donnai-Barrow Syndrome

- a.k.a. Faciooculoacousticorenal syndrome (FOAR)

Main Features

- Facial abnormalities
 - Prominent Brow
 - Short Nose
 - Hypertelorism
- Ocular anomalies
 - Myopia (often high)
 - Iris hypoplasia
 - Retinal Detachment
- Acoustic problems
 - sensorineural hearing loss
- Renal problems
 - Proteinuria

Major Ocular Features

- Hypertelorism
- High myopia
- Loss of vision
- Iris coloboma
- Iris hypoplasia
- Cataract
- Enlarged globes and prominent eyes
- Downslanting palpebral fissures
- Underorbital skin creases
- Retinal detachment
- Retinal dystrophy

Other Features

- Diaphragmatic hernia
- Omphalocele
- Macrocephaly
- Ventricular septal defect
- Corpus callosum partial or complete agenesis

Cause

- Mutation in the Low density lipoprotein receptor related protein 2 gene (LRP2)

Resources

- [LRP2 Gene review](#)
- [OMIM for Donnai-Barrow Syndrome](#)

[syndrome](#)

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