

Congenital Disorders of the Optic Nerve

Excavations and Hypoplasia

Optic Disc Coloboma

- Abnormal fusion of the optic cup
- Sporadic or inherited

Associations

- CHARGE association (coloboma, choanal atresia, congenital heart disease, and multiple other abnormalities)
- Walker- Warburg syndrome
- Goltz focal dermal hypoplasia
- [Aicardi syndrome](#)
- [Goldenhar syndrome](#)
- Linear sebaceous naevus syndrome
- [Dandy Walker malformation](#)
- Renal coloboma syndrome (with a mutation of PAX2 transcription)

Morning Glory Anomaly

- congenital optic disc dysplasia

Associations

- Typically isolated
- transphenoidal encephalocele and hypopituitarism
- Retinal detachment
- Subretinal neovascularization

Peripapillary staphyloma

- disturbance of peripapillary scleral development

Associations

- Usually isolated and unilateral

- frontonasal displasia with encephalocele
- facial capillary hemangioma
- linear nevus sebaceous syndrome

Optic Nerve Hypoplasia

see entry [Optic Nerve Hypoplasia](#)

Optic Disc Cupping in Periventricular Leukomalacia

- disturbance of development of the white matter during intrauterine development can lead to optic disc cupping and thinning of the neuroretinal rim
- May be the result of trans-synaptic degeneration

References

- [Congenital disorders of the optic nerve: excavations and hypoplasia. Dutton GN. Eye 2004](#)

From:
<https://childreneye.org/wiki/> - **Children's Eye Wiki**

Permanent link:
https://childreneye.org/wiki/doku.php?id=congenital_disorders_optic_nerve

Last update: **2025/01/10 16:13**

