CONGENITAL EYE DISORDERS
CONGENITAL ABNORMALITIES OF GLOBE

- **ANOPHTHALMOS**: total absence of ocular tissues (failure of formation of the optic pit)
- **CYSTIC EYE**: total absence of ocular tissues (after formation of the optic pit)
- **CRYPTOPHTHALMOS**: failure of differentiation of eyelid structures.
- **MICROPHTHALMOS**: small eye with major internal disorganization.
- **NANOPHTHALMOS**: small eye without major internal disorganization.
ANOPHTHALMOS

Figure 6-5  Bilateral clinical anophthalmos. Eyelids, brows, and orbits are well formed but small.

Figure 6-6  Partially opened small eyelids of anophthalmic orbit.
Figure 6-10 Orbital cysts replacing both globes.
Figure 6-11  A. Complete cryptophthalmos, both eyes. B. Incomplete cryptophthalmos of right eye with eyelid fused to cornea superonasally.
MICROPHTHHALMOS

Figure 6-12  Severe microcornea and microphthalmos OD. Both irides are colobomatous.
CONGENITAL ABNORMALITIES OF LID

- Congenital ptosis
- Coloboma
- Belpharophimisis
- Epicanthus
- Ancylobelpharon
- Epibelpharon
Congenital ptosis

**Simple congenital ptosis**

- Developmental dystrophy of levator muscle
- Occasionally associated with weakness of superior rectus

Unilateral or bilateral ptosis of varying severity

In downgaze ptotic eyelid is slightly higher

Frequent absence of upper lid crease

Usually poor levator function
Congenital ptosis

FIG XII-13—Bilateral asymmetric congenital ptosis. A, Note margin–reflex distance (MRD = 5.0 mm OD, 1.0 mm OS). Normal = 4.5 mm. B, Up-gaze accentuates ptosis. C, Down-gaze exhibits eyelid lag.
Coloboma

FIG XI-4—True coloboma of upper eyelid.
Belpharophimosis

• This congenital eyelid syndrome is an autosomal dominantly inherited blepharophimosis usually presenting with telecanthus (widened intercanthal distance), epicanthus inversus (fold of skin extending from the lower to upper eyelid), and severe ptosis.
FIG XI-1—Blepharophimosis syndrome.
Blepharophimosis syndrome

- Rare congenital disorder
- Dominant inheritance

- Moderate to severe symmetrical ptosis
- Short horizontal palpebral aperture
- Telecanthus (lateral displacement of medial canthus)
- Epicanthus inversus (lower lid fold larger than upper)
- Lateral inferior ectropion
- Poorly developed nasal bridge and hypoplasia of superior orbital rims
Epicanthus

- Epicanthus is a medial canthal fold that may result from immature midfacial bones or a fold of skin and subcutaneous tissue.
- The condition is usually bilateral.
- Four types of epicanthus are described:
  - *Epicanthus tarsa/is* if the fold is most prominent in the upper eyelid.
  - *Epicanthus in versus* if the fold is most prominent in the lower eyelid.
  - *Epicanthus palpebra/is* if the fold is equally distributed in the upper and lower eyelids.
  - *Epicanthus supracci/iaris* if the fold arises from the eyebrow region running to the lacrimal sac.
Figure 16-6  Epicanthus, bilateral. Top, Epicanthus tarsalis. Bottom, Epicanthus palpebralis.