Incontientia Pigmenti

(Block-Sulzberger Syndrome)

Main Features

- Involves skin, brain and eyes
- X-Linked dominant, usually lethal to males
- Skin normal at birth but erythema and bullae develop in first few days of life usually on extremities
- Verrucous changes at two months of age then small hyperpigmented macules on trunk appear

Eye Findings

- 25-33%
- Proliferative retinal vasculopathy that resembles retinopathy of prematurity
- At birth incomplete peripheral vascularization develops into abnormal vascular shunts and neovascular membranes
- Retinal detachment often develops
- Retrolental membrane formation (pseudoglioma)
- Microphthalmos, cataract, glaucoma, optic atrophy, strabismus, nystagmus
- Treated like retinopathy of prematurity

Etiology

Caused by mutations in the NEMO gene Xq28

Other Findings

- CNS abnormalities (33%)
  - microcephaly, hydrocephalus,seizures, mental deficiency
- Dental abnormalities (66%)
  - missing or malformed teeth
- Less common: scoliosis, skull deformities, cleft palate, dwarfism

References

- Catalano RA. Incontientia pigmenti.AM J Ophthalmol. 1990;110:696-700
- OMIM

syndrome