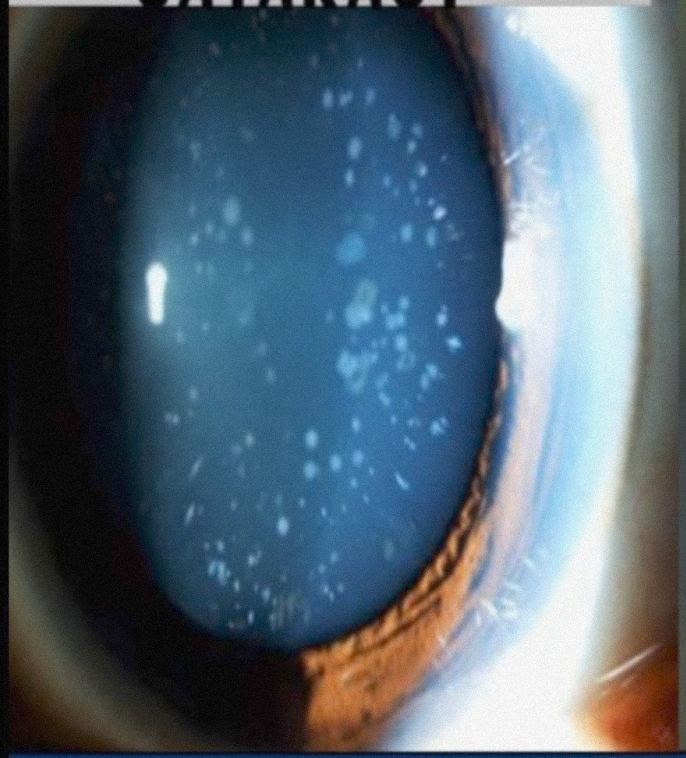
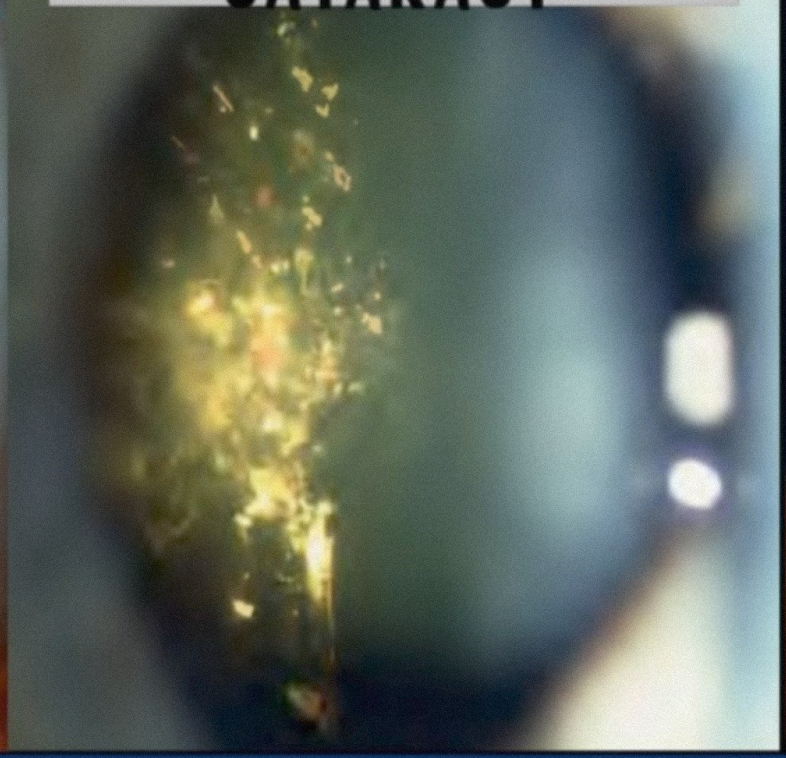
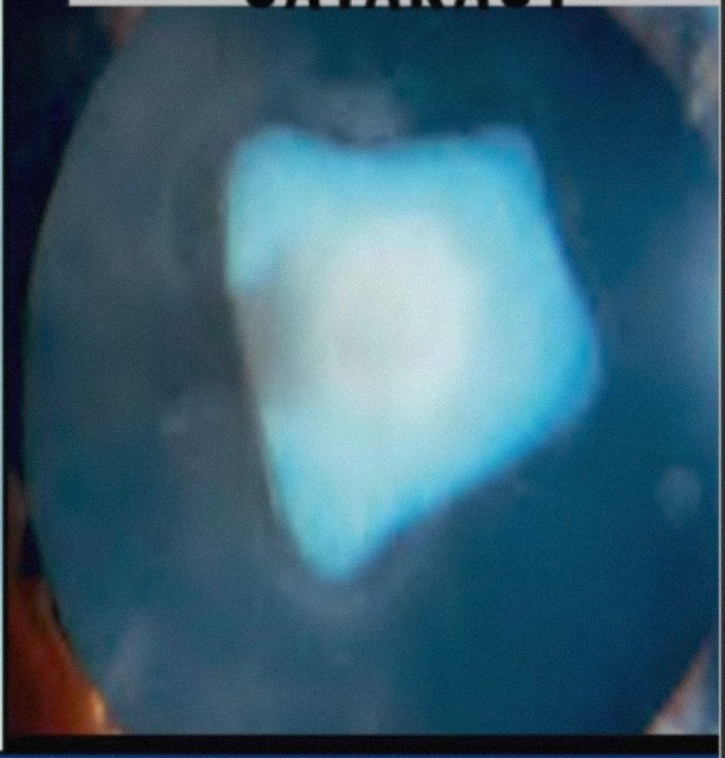
BLUE DOT CATARACT



CHRISTMAS CATARACT



SHIELD/ATOPIC CATARACT



SYNDROMIC CATARACTS

MUSCULOSKELETAL SYNDROMES

CRANIOFACIAL ANOMALIES

CHROMOSOMAL DISORDERS

DENTAL DISORDERS

RENAL SYNDROMES

SKIN DISORDERS

MUSCULOSKELETAL SYNDROMES

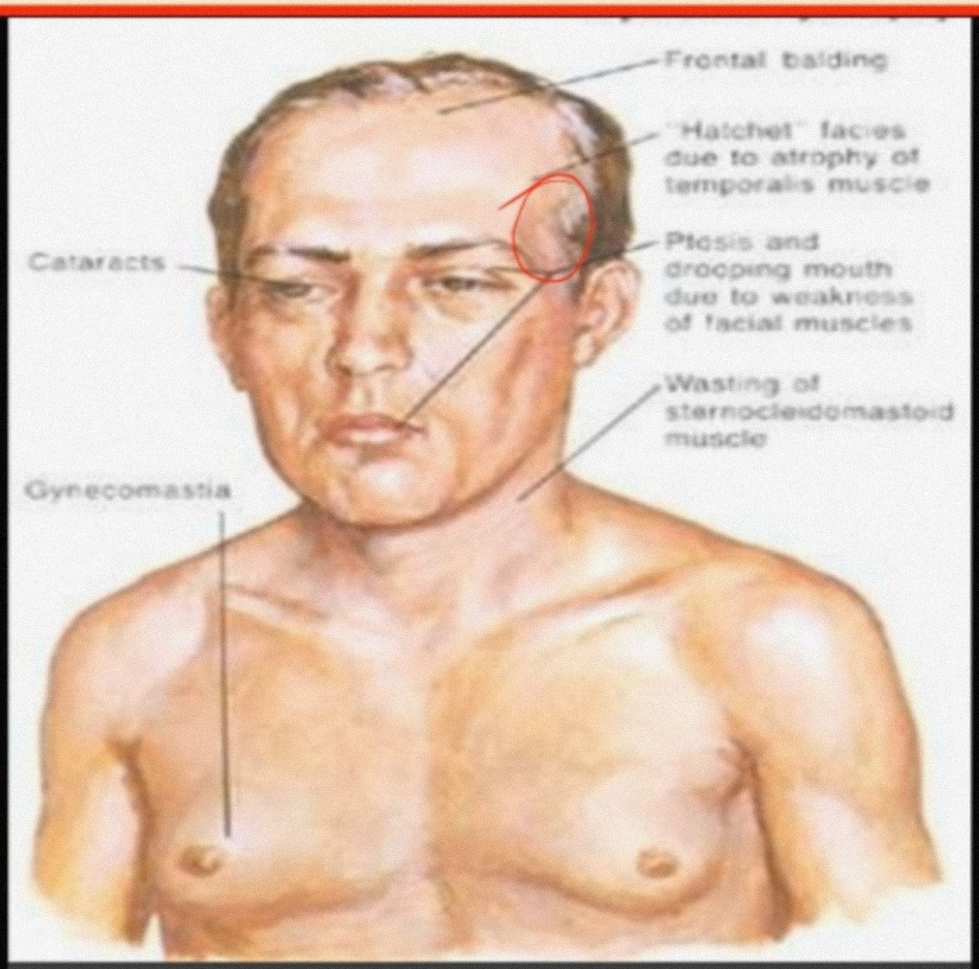
SMITH LEMIS OPTIZ SYNDROME

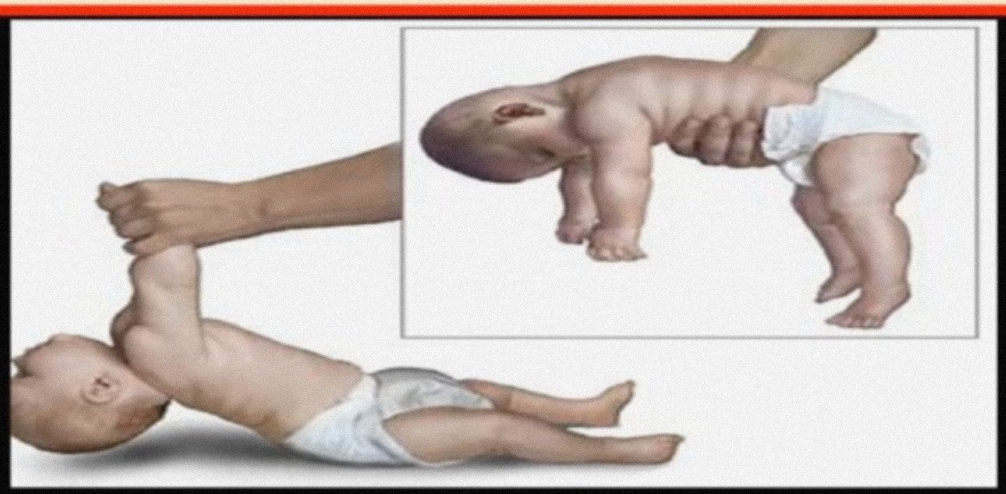
CONRADI HUNERMANN SYNDROME

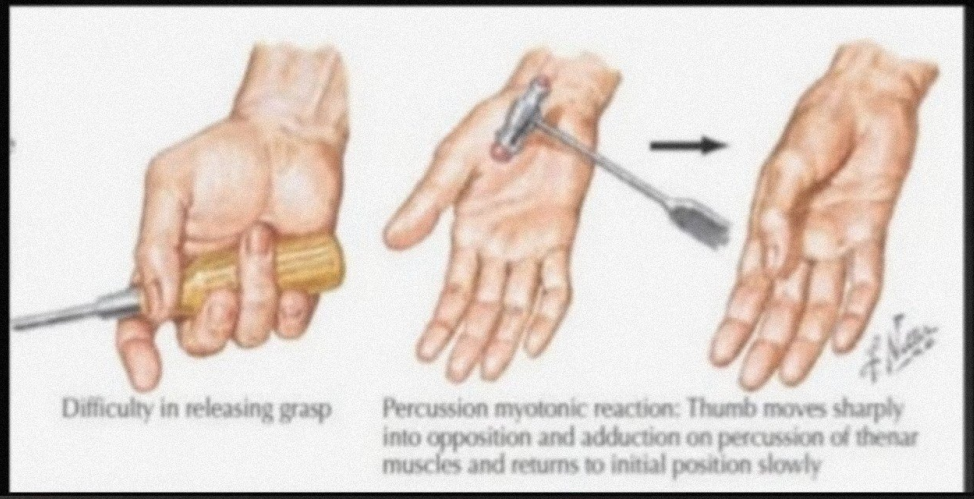
MYOTONIC DYSTROPHY

WEIL MARCHESANI SYNDROME

MYOTONIC DYSTROPHY



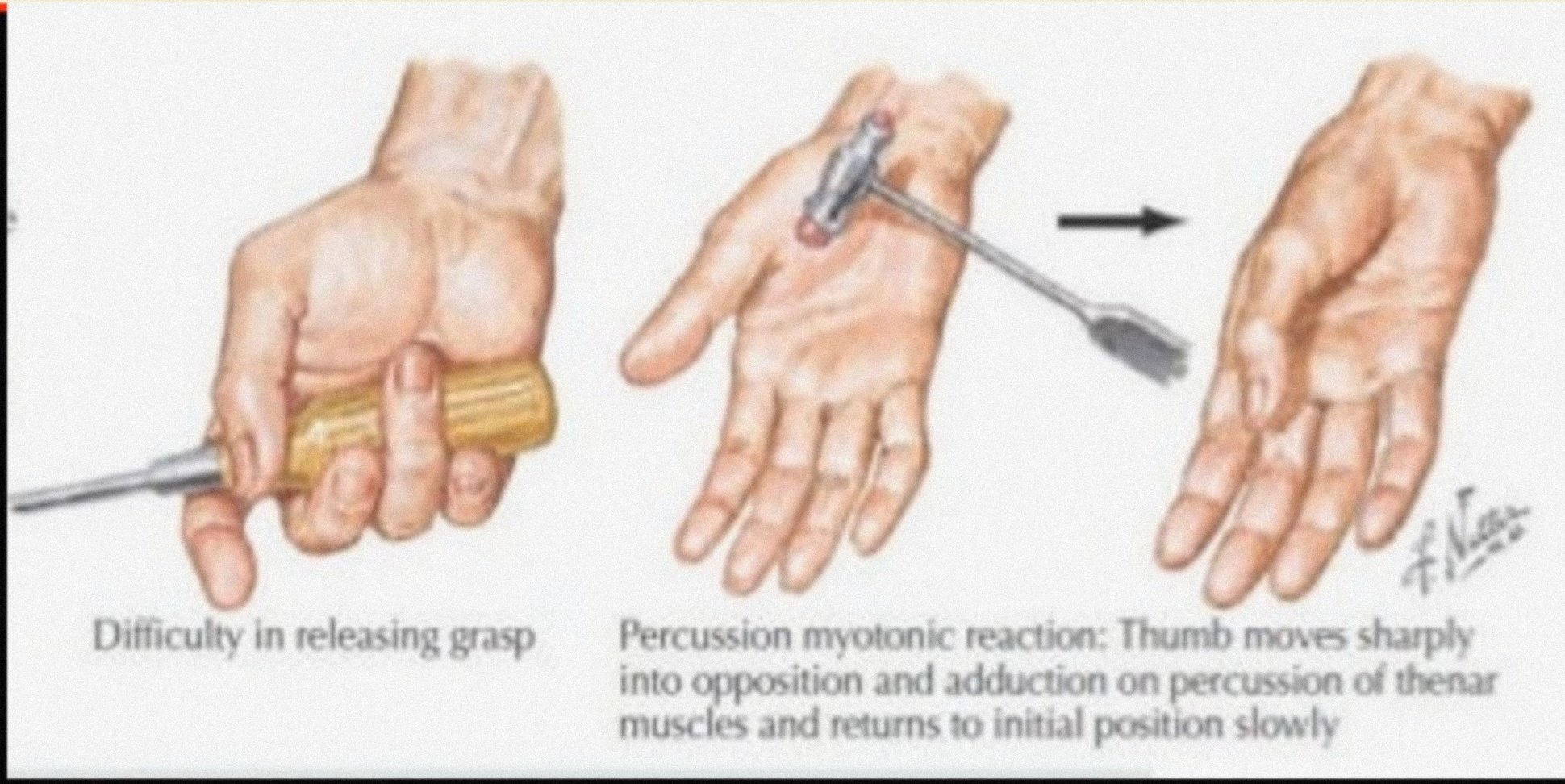




Difficulty in releasing grasp

Percussion myotonic reaction: Thumb moves sharply into opposition and adduction on percussion of thenar muscles and returns to initial position slowly

MYOTONIC DYSTROPHY

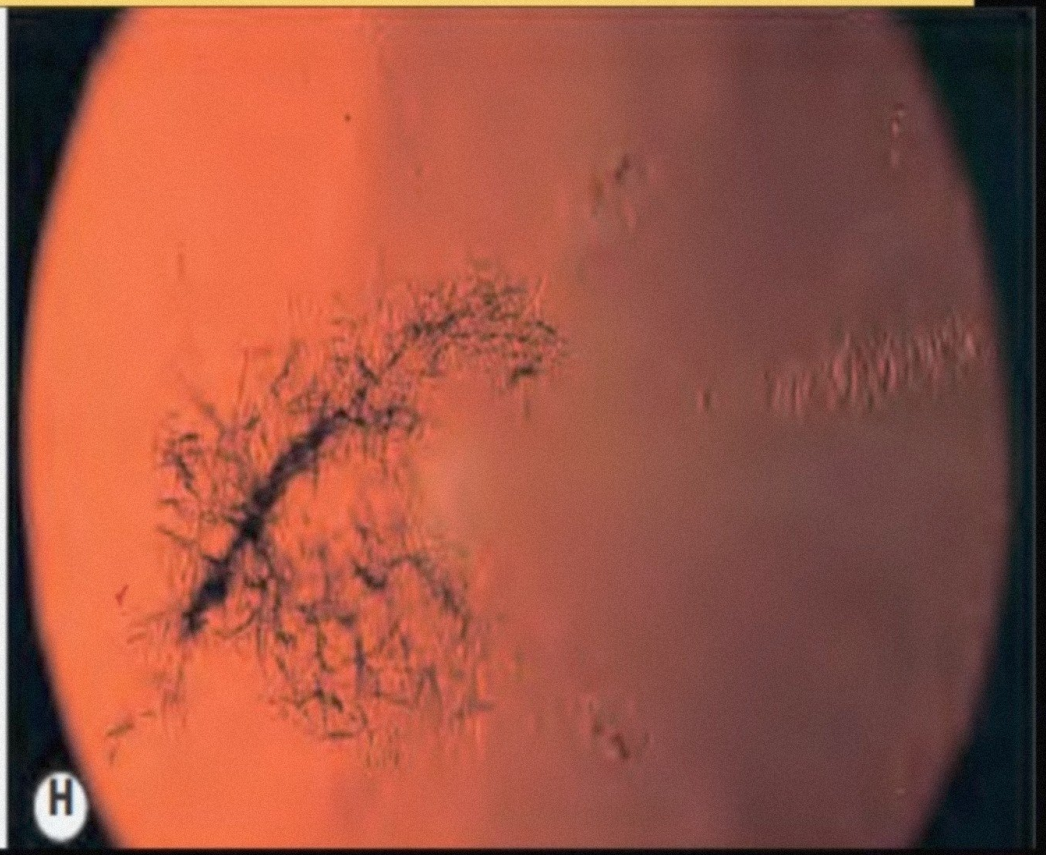


Difficulty in releasing grasp

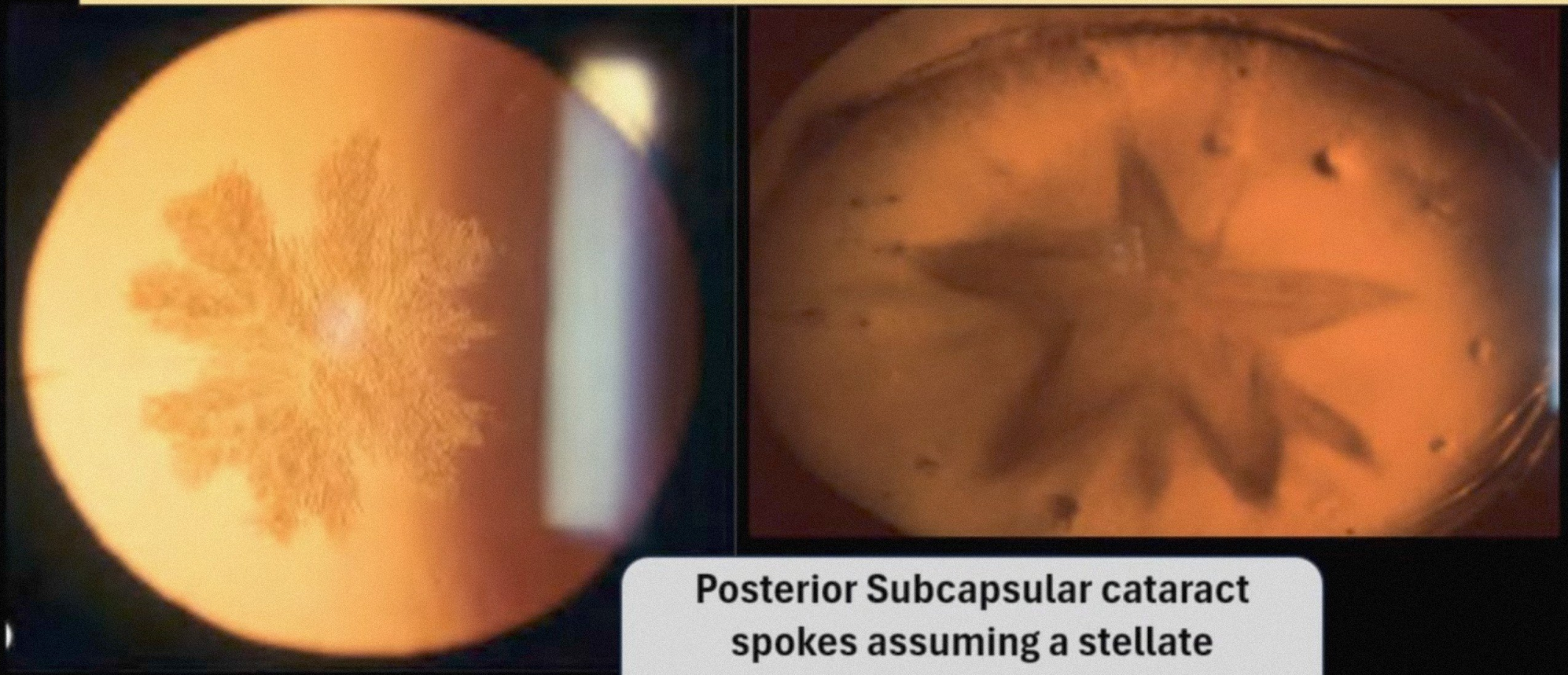
Percussion myotonic reaction: Thumb moves sharply into opposition and adduction on percussion of thenar muscles and returns to initial position slowly

MYOTONIC DYSTROPHY

CHRISTMAS TREE CATARACT



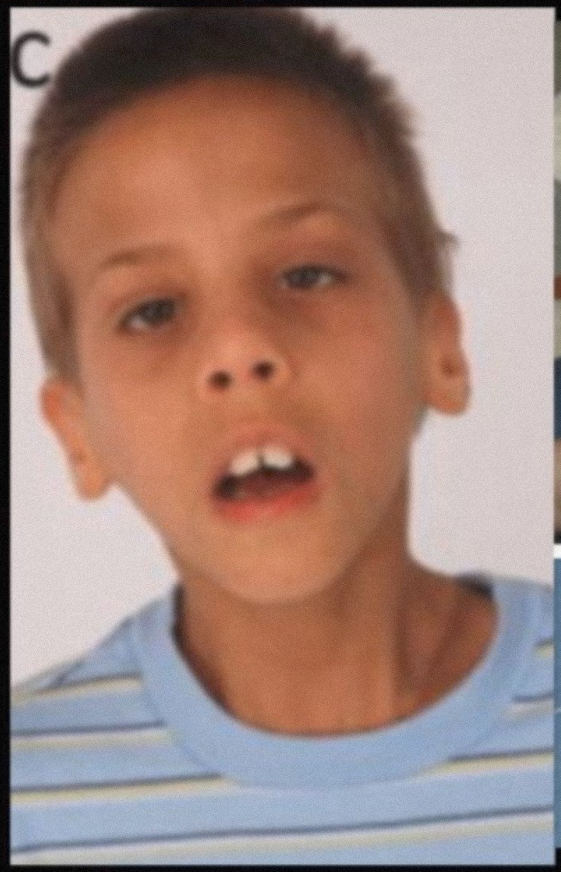
MYOTONIC DYSTROPHY

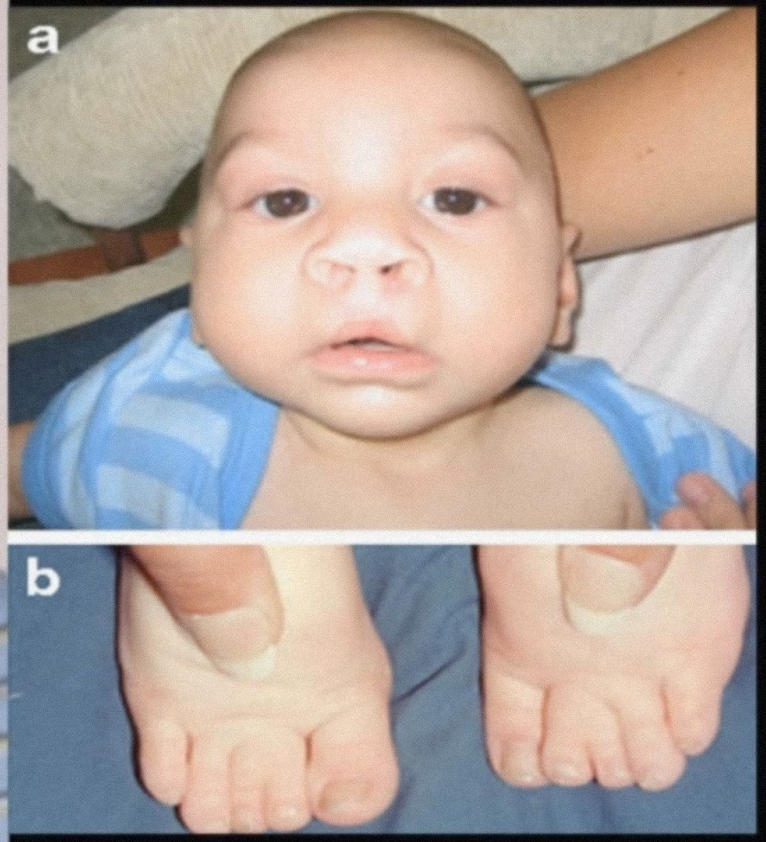


Posterior Subcapsular cataract spokes assuming a stellate morphology !

SMITH LEMLI OPITZ SYNDROME

- Microcephaly with bitemporal narrowing
- A short upturned nose with **ANTEVERTED NARES**
- **LONG PHILTRUM**
- Unilateral or bilateral ptosis, epicanthus.
- Retrognathia
- Polydactyly
- Syndactyly
- Short stature

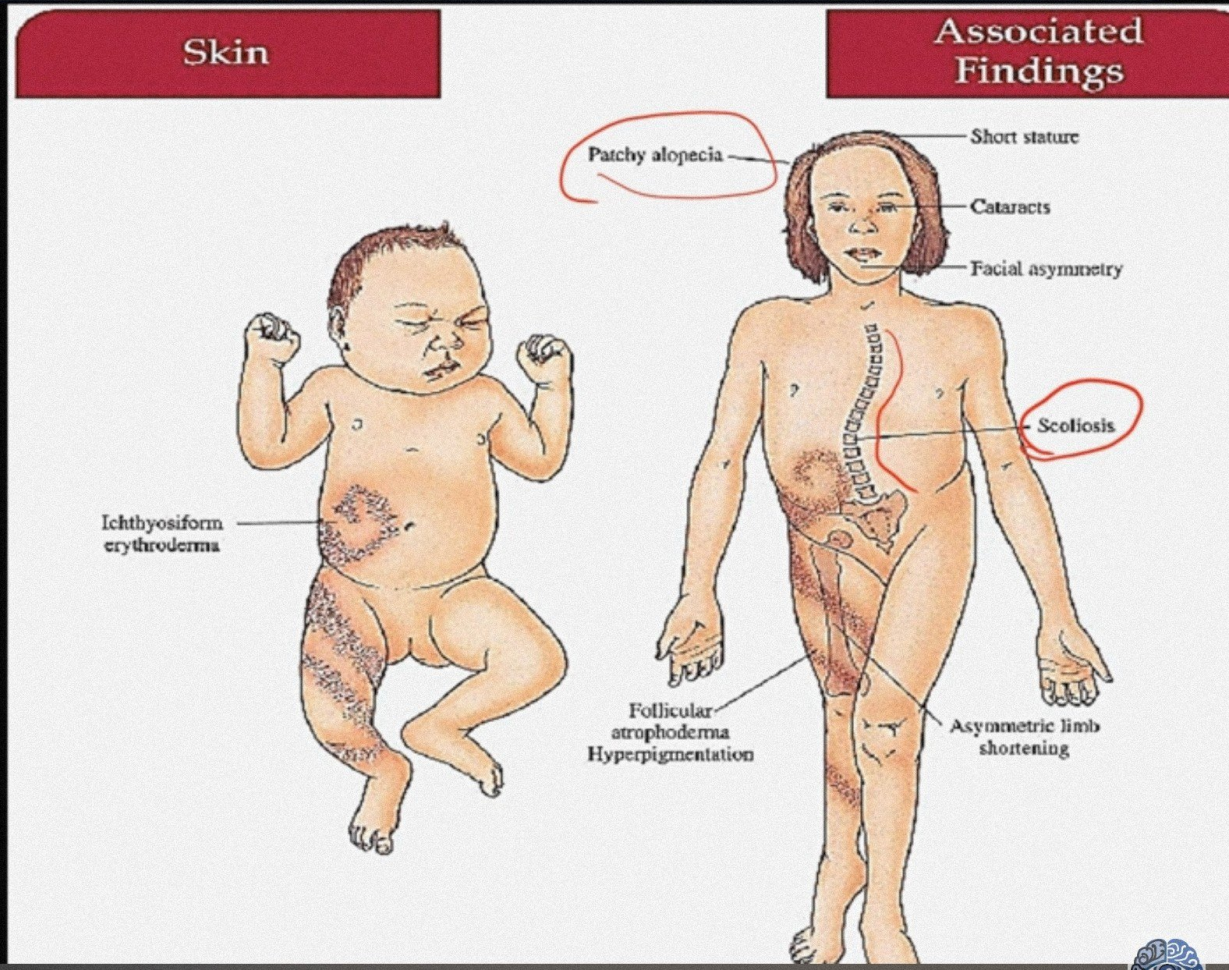




CONRADI HUNERMANN SYNDROME

- Short stature
- Patchy alopecia
- **Scoliosis**
- **Asymmetric limb shortening**

- **MICROPHTHALMOS**
  MICROCORNEA
  CATARACTS

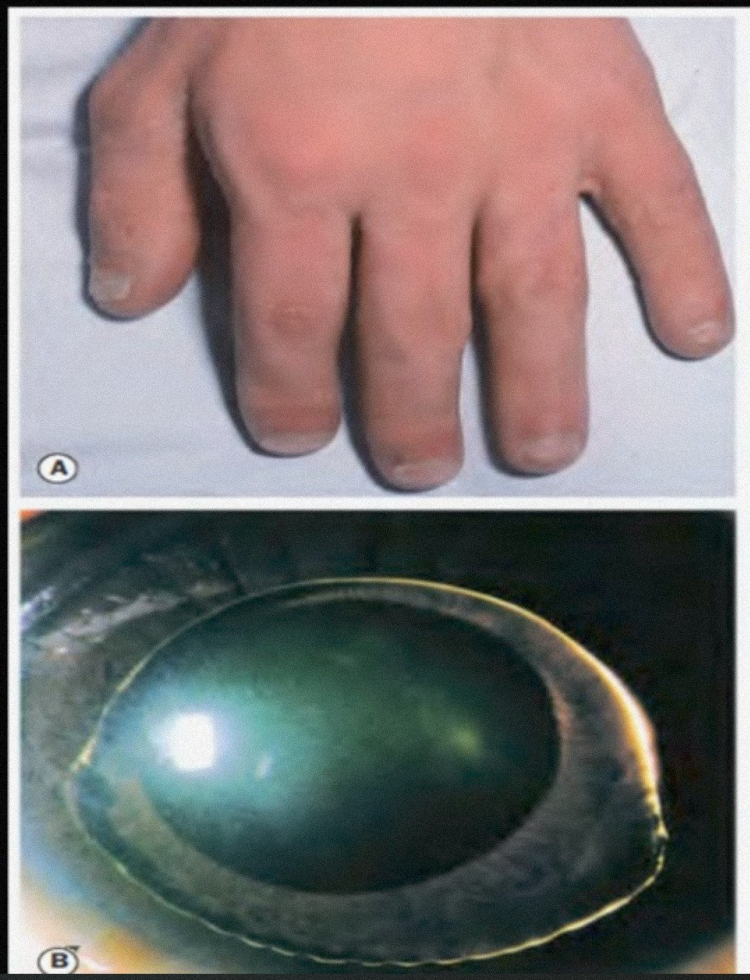


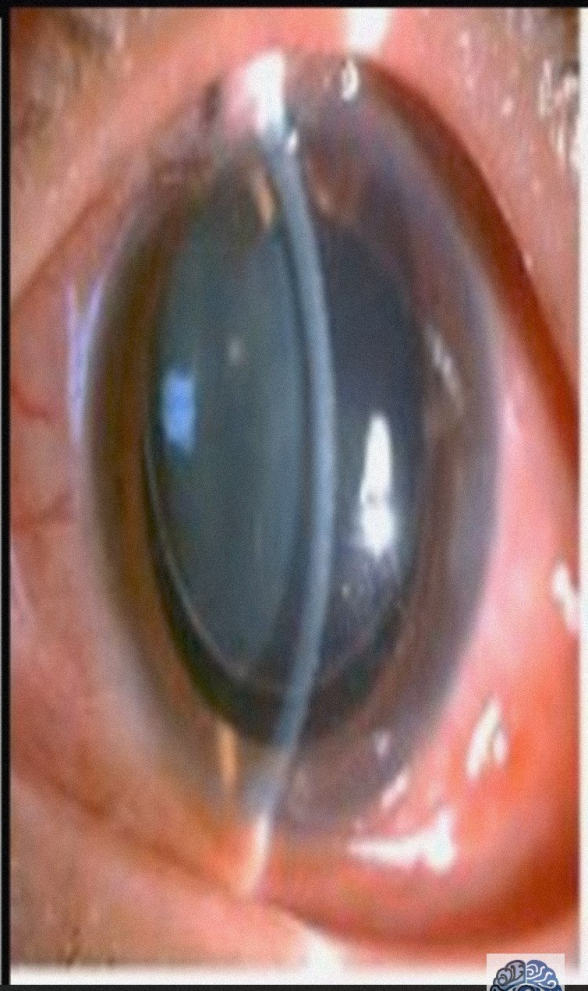
WEIL MARCHESANI SYNDROME

SPHEROPHAKIA-BRACHYMORPHIA SYNDROME

- Stocky build and have small, stubby fingers.
- Bradydactly

- Microspherophakia
- Anterior dislocation of lens





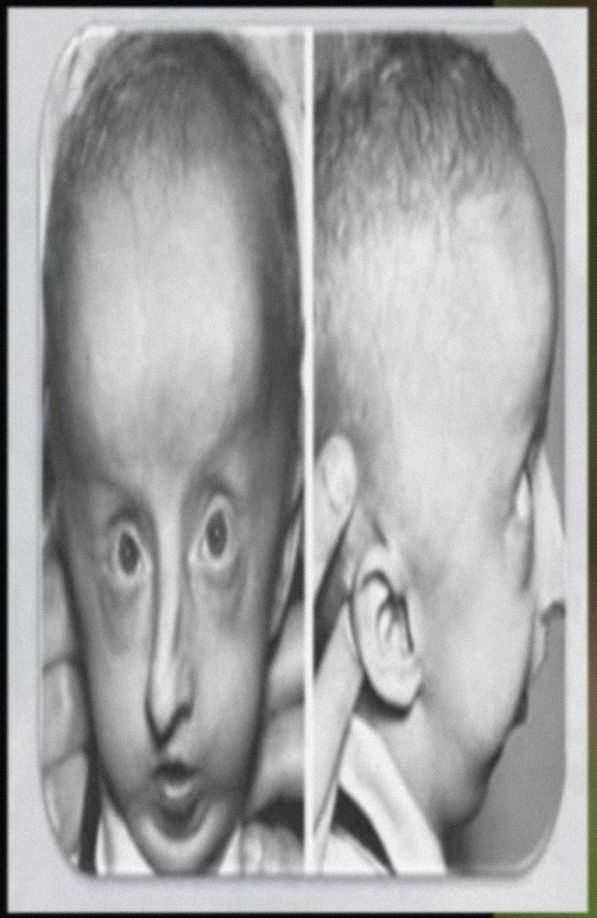
HALLERMAN STRIEF FRANCOID SYNDROME

CRANIOFACIAL ANOMALIES AND CATARACT

RUBINSTEIN TAYBI

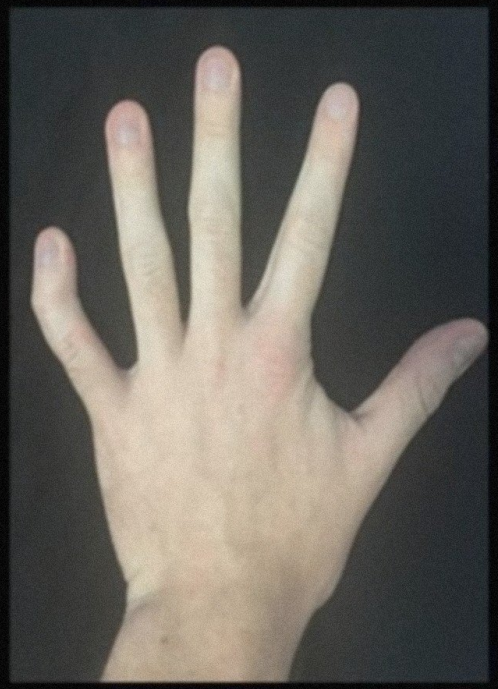
HALLERMAN STRIEF FRANCOID SYNDROME

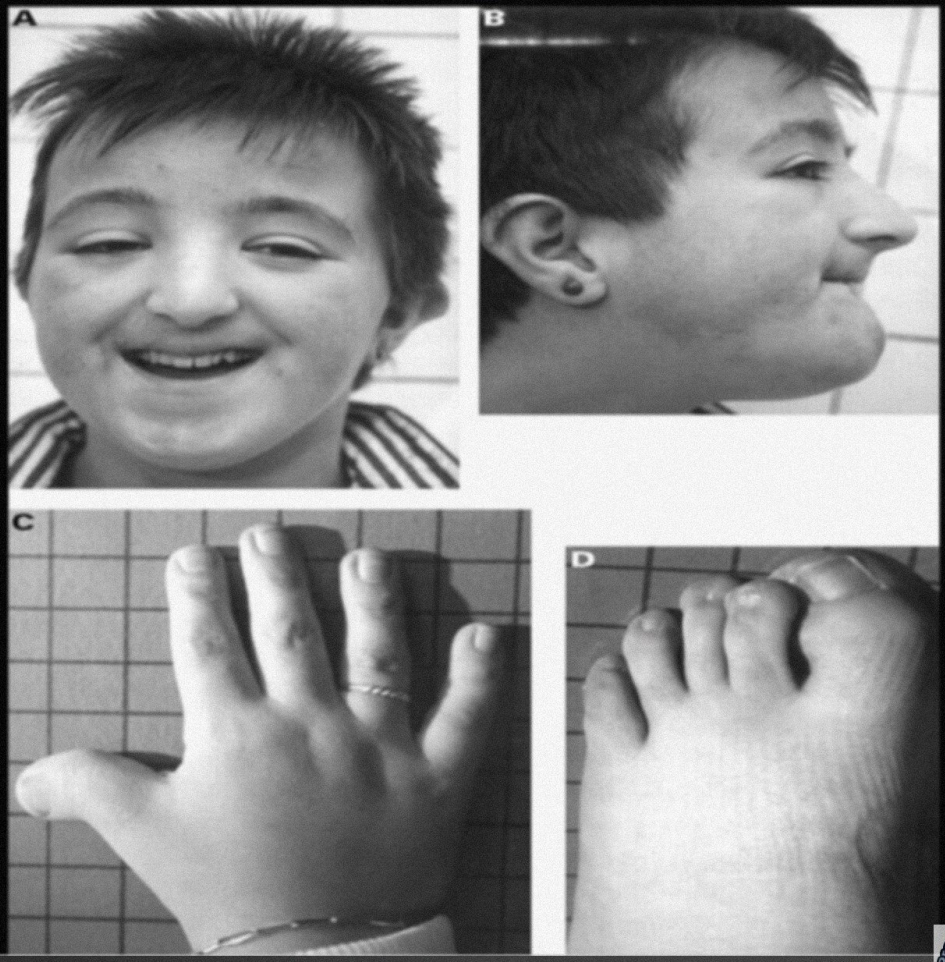
- Abnormal facial appearance
- **Bird Like Facies**
- Dental abnormalities,
- Hypotrichosis
- Skin atrophy
- Proportionate short stature
- Ophthalmic features including **microphthalmia** and congenital bilateral cataracts.



RUBINSTEIN TAYBI SYNDROME

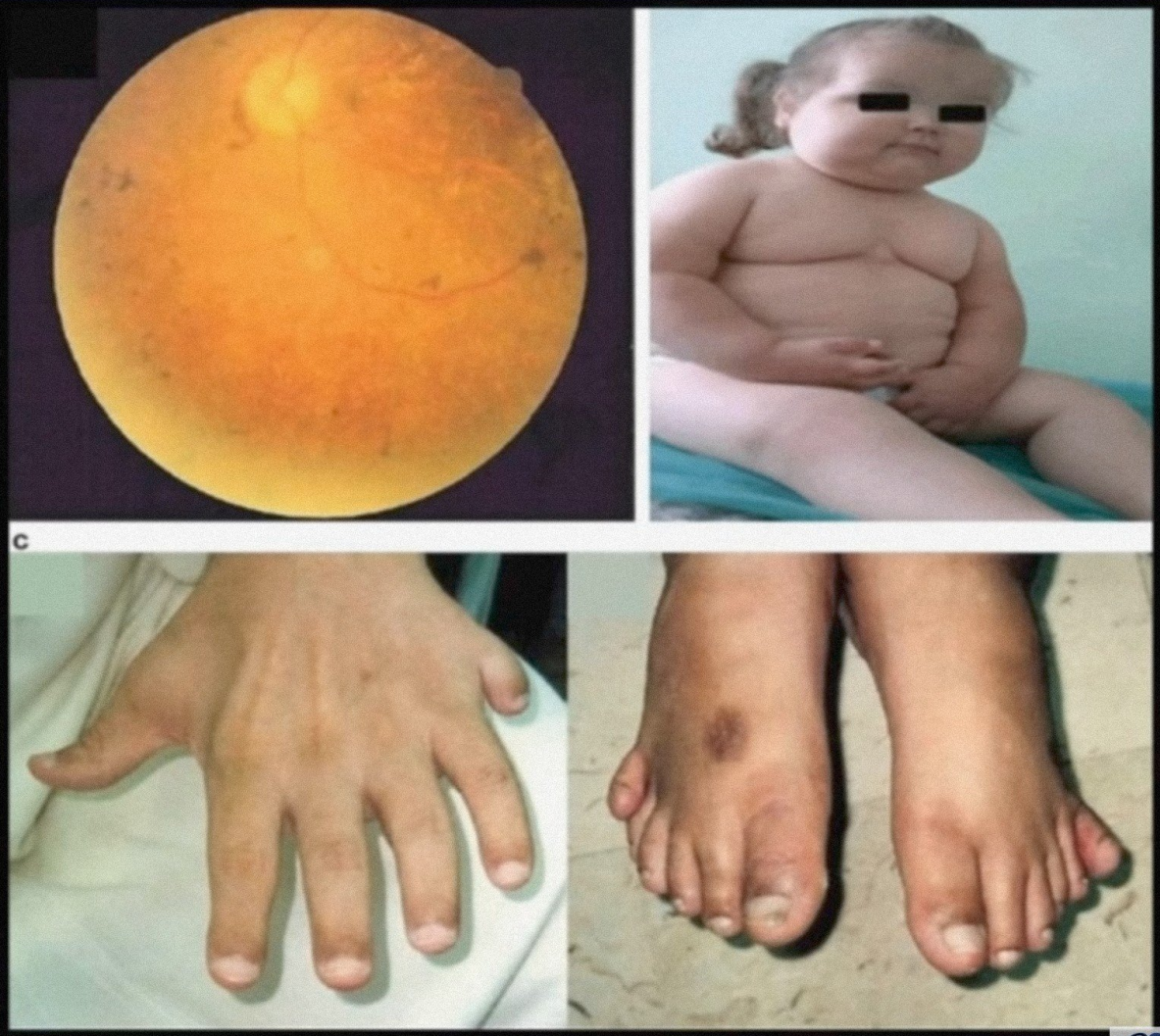
- Broad first toe
- Broad thumb
- Clinodactyly
- Microcephaly



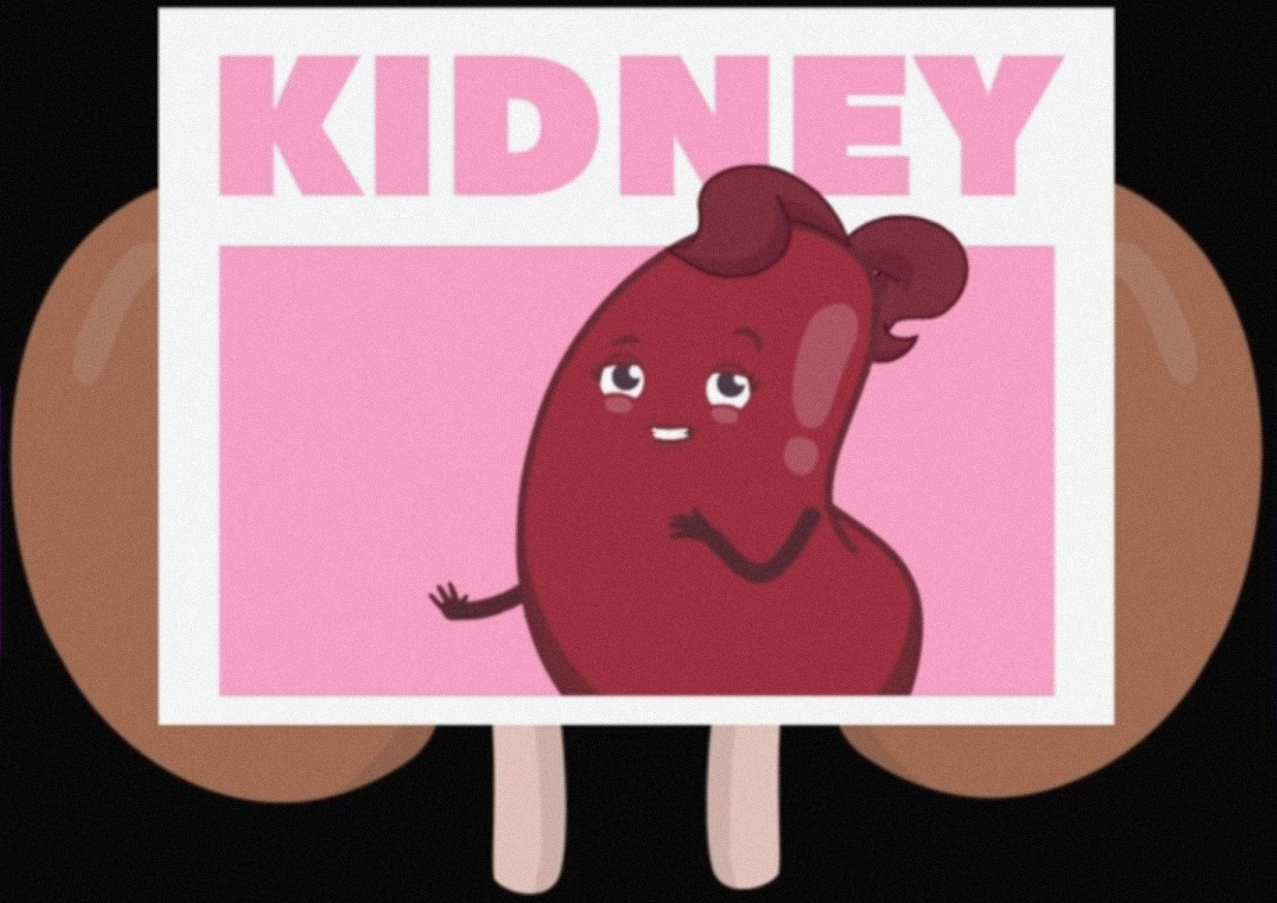


BARDET BEIDEL SYNDROME

- Retinal degeneration
- Truncal obesity
- Cognitive impairment
- **Postaxial polydactyly**
- Hypogonadism/genitourinary anomalies
- Renal abnormalities

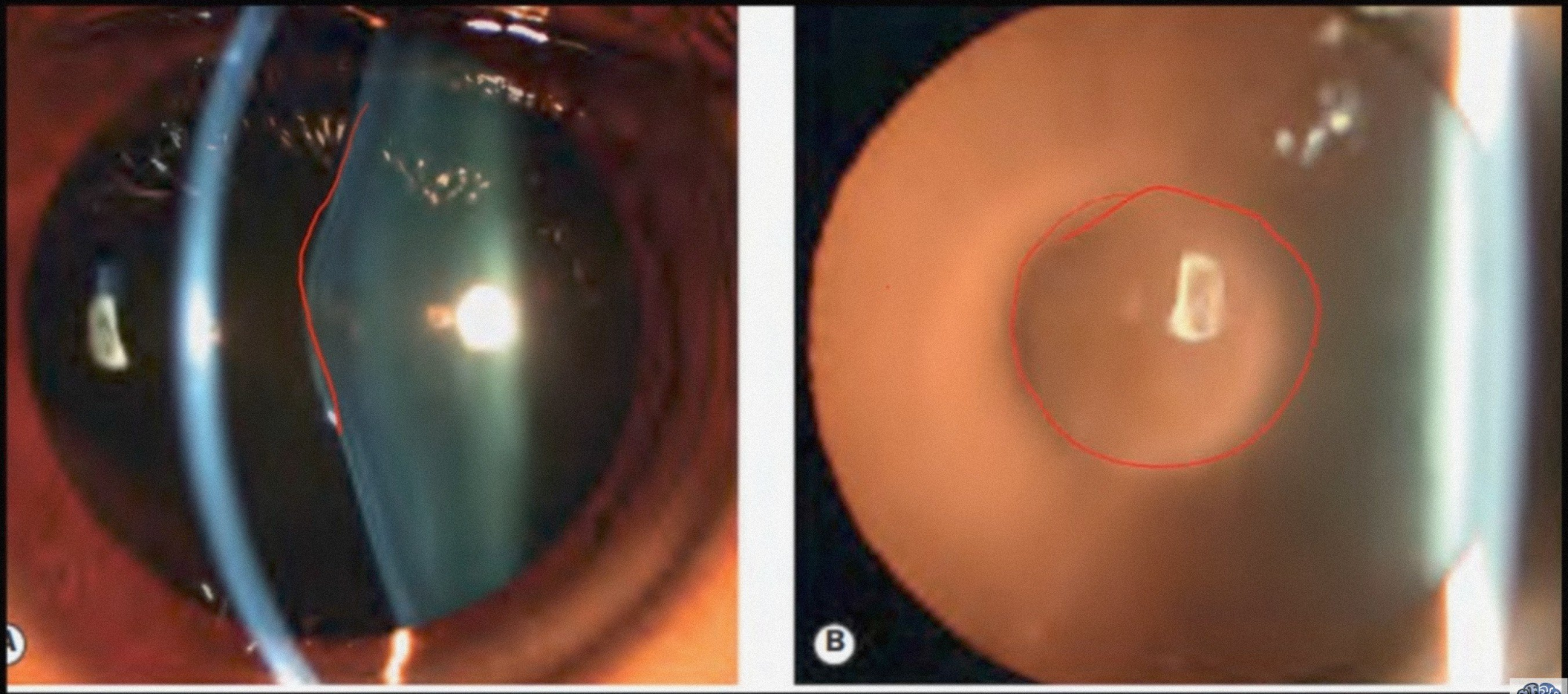


ALPORT SYNDROME

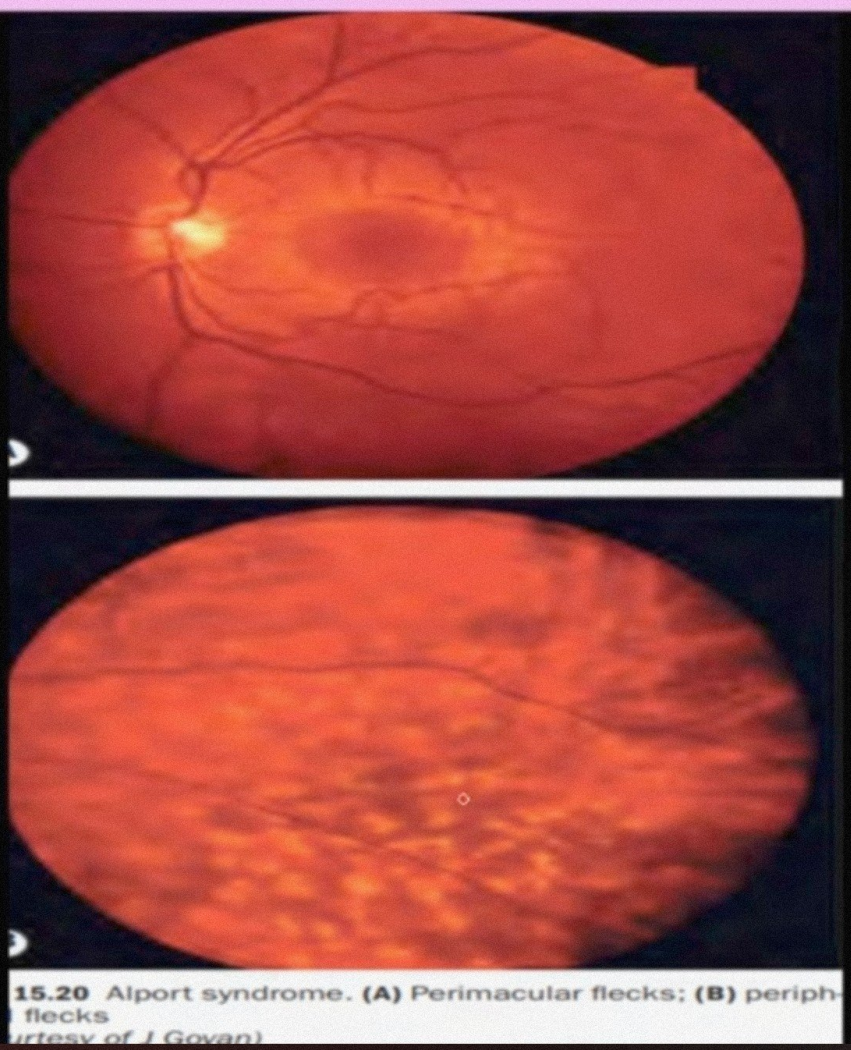


LOWE SYNDROME

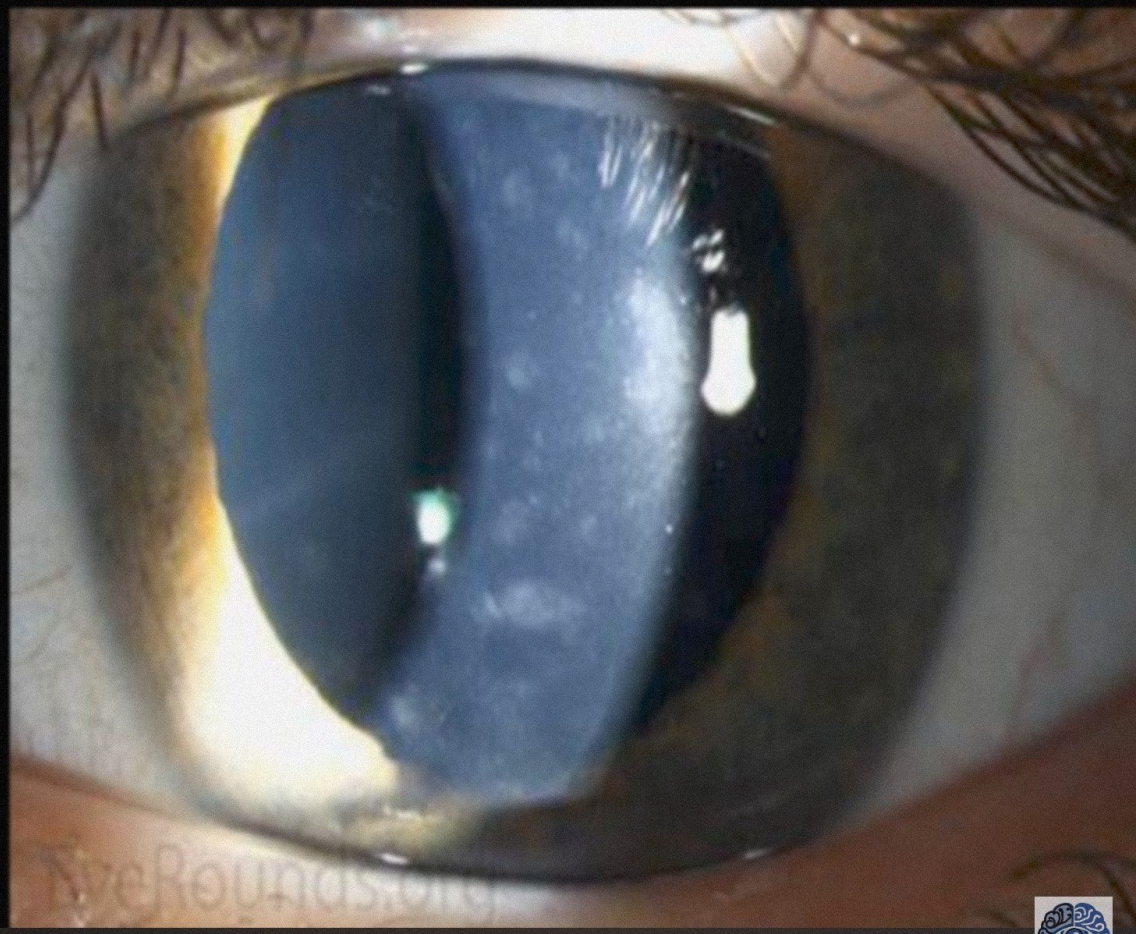
ALPORT SYNDROME



ALPORT SYNDROME



15.20 Alport syndrome. **(A)** Perimacular flecks; **(B)** periph-
flecks
urtesy of J Govan)



LOWE SYNDROME

- Oculocerebrorenal syndrome of Lowe (OCRL)
- Male children
- Bilateral congenital cataracts
- Associated with **LENTICONUS**
- Severe hypotonia
- Proteinuria may be the most sensitive marker for renal involvement of LS.
- Fanconi syndrome

T : TOXOPLASMA

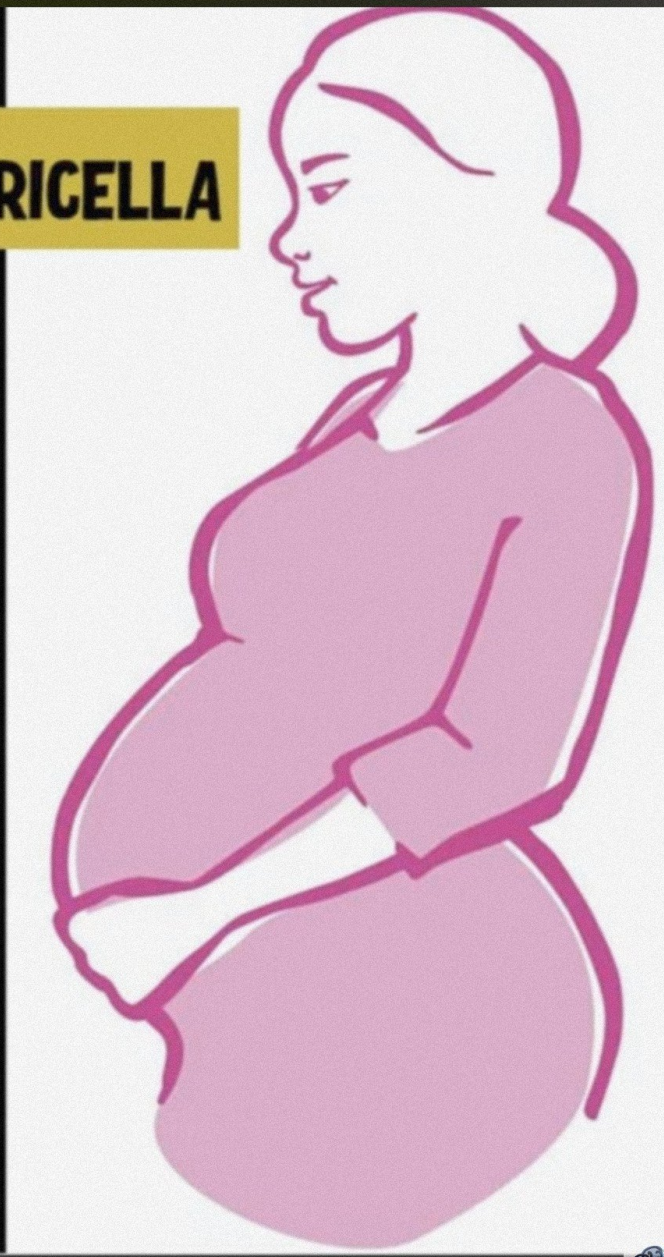
OTHER PATHOGENS (syphilis)

RUBELLA

CYTOMEGALOVIRUS

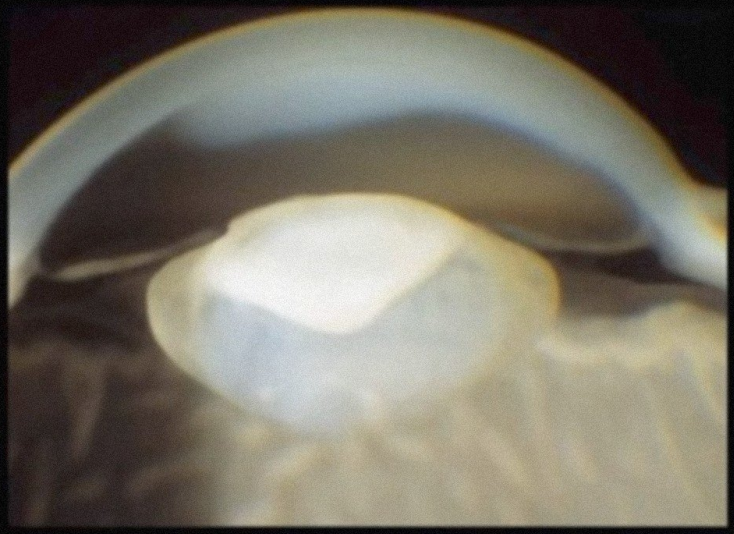
HERPES SIMPLEX VIRUS

VARICELLA

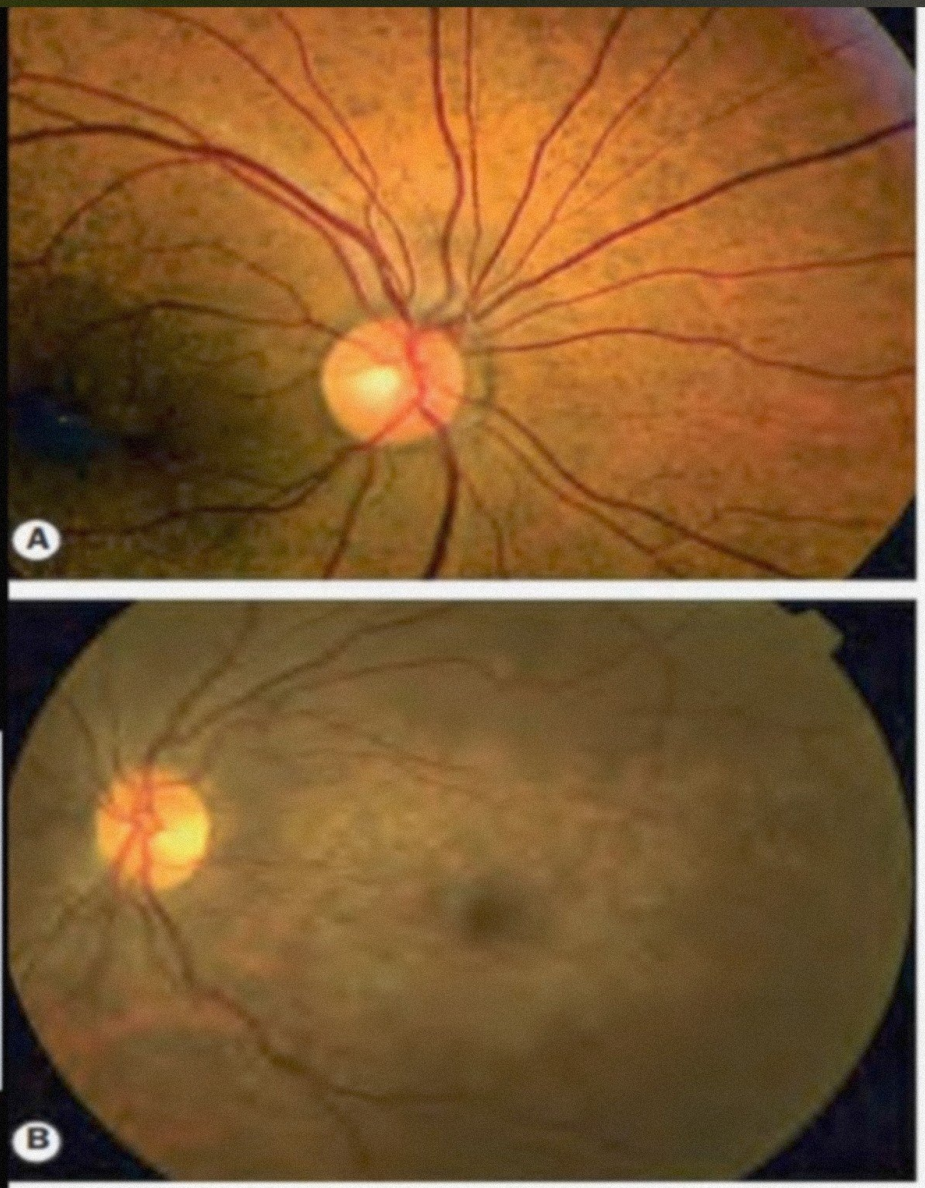


RUBELLA CATARACT

- Pearly nuclear or more diffuse unilateral or bilateral cataract occurs in around 15%.



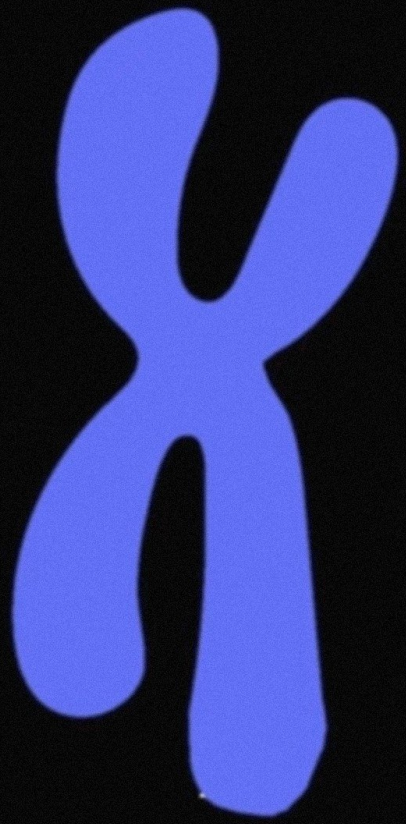
SALT and pepper retinopathy



g. 12.56 Rubella retinopathy. **(A)** 'Salt and pepper' appear-
nce; **(B)** predominantly manifesting in the macula

TRISOMY 21 / DOWNS SYNDROME

TRISOMY 13 / PATAU SYNDROME

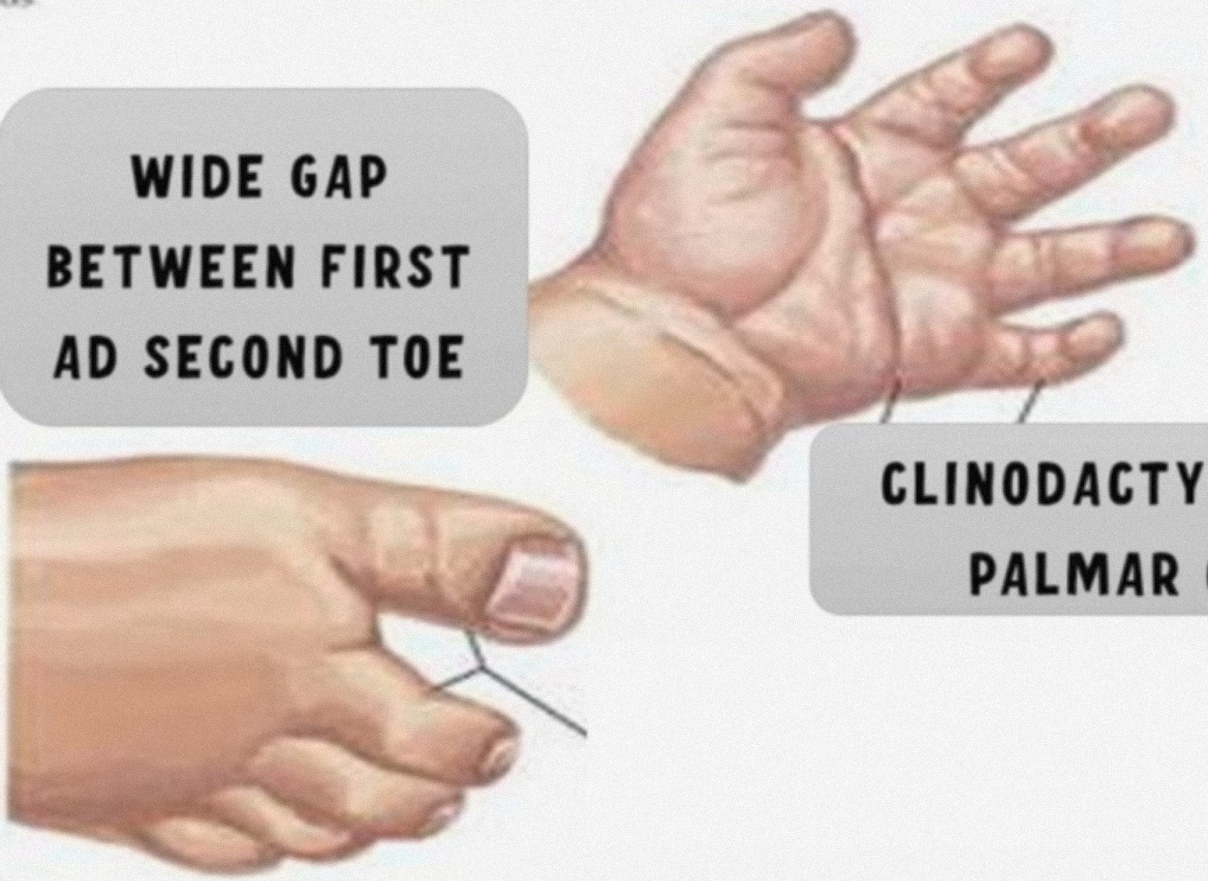


TRISOMY 18 / EDWARDS SYNDROME

CRI-DU-CHAT SYNDROME

TRISOMY 21 / DOWNS SYNDROME

WIDE GAP
BETWEEN FIRST
AD SECOND TOE



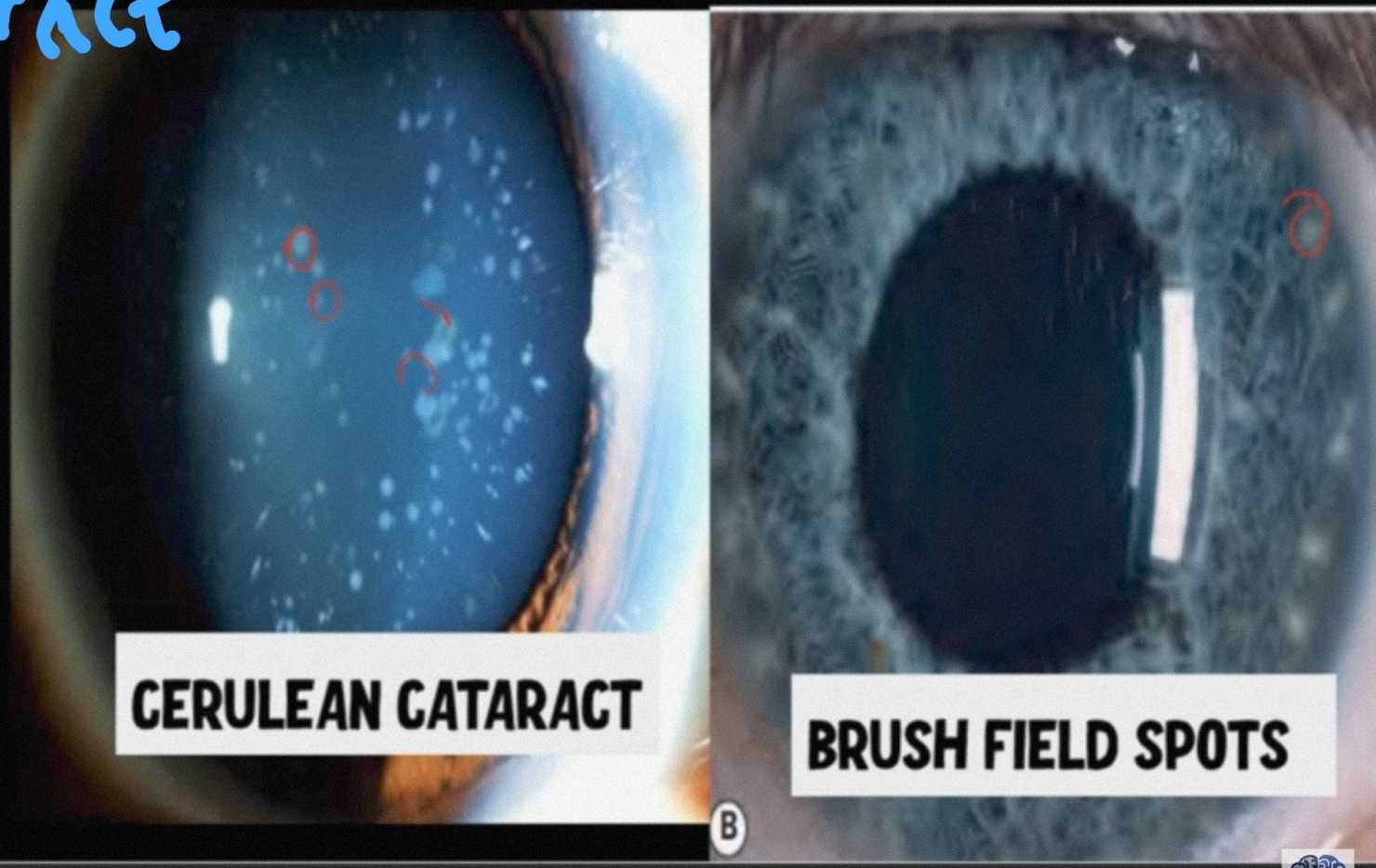
CLINODACTYLY . SINGLE
PALMAR CREASE

EPICANTHUS.
UPWARD SLANT.
Flat nasal bridge

TRISOMY 21 / DOWNS SYNDROME

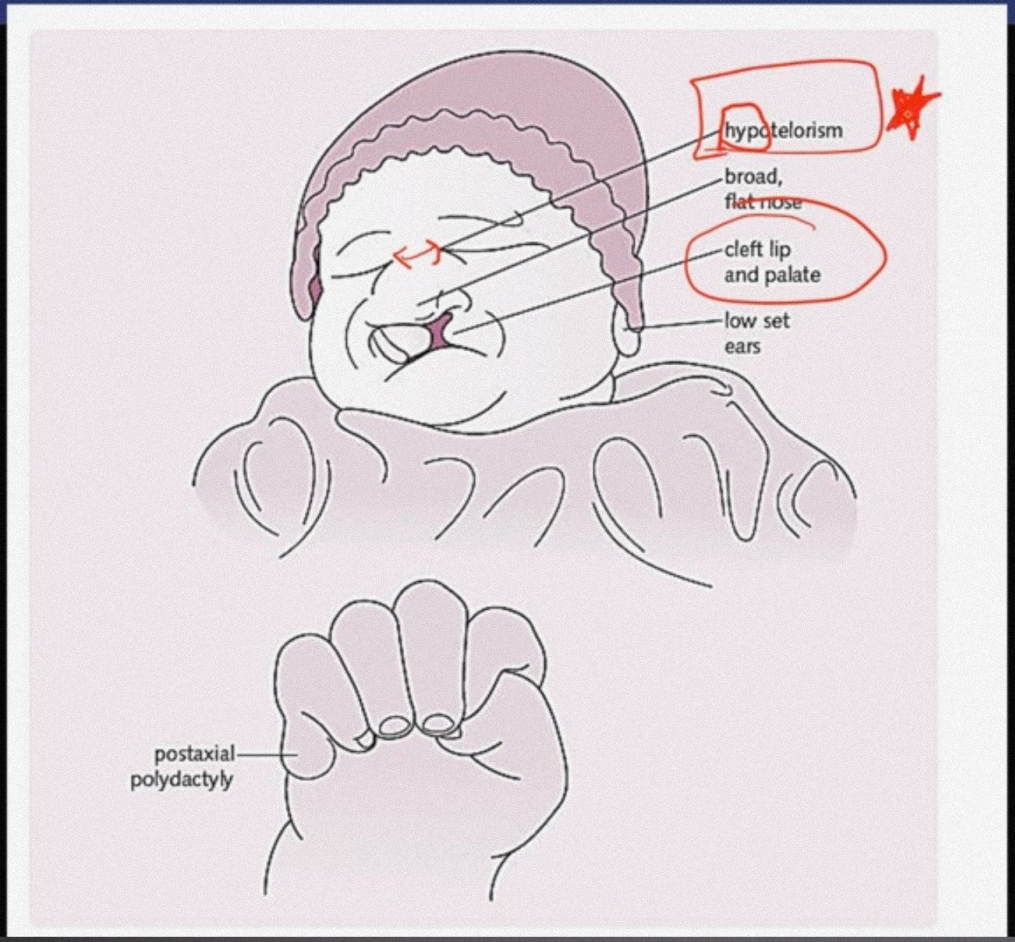
Blue Dot Cataract

- Cataract of varied morphology (75%).
- Symmetrical opacities and often develop in late childhood.
- Other features include iris Brush field spots
- Keratoconus
- Glaucoma



TRISOMY 13 / PATAU SYNDROME

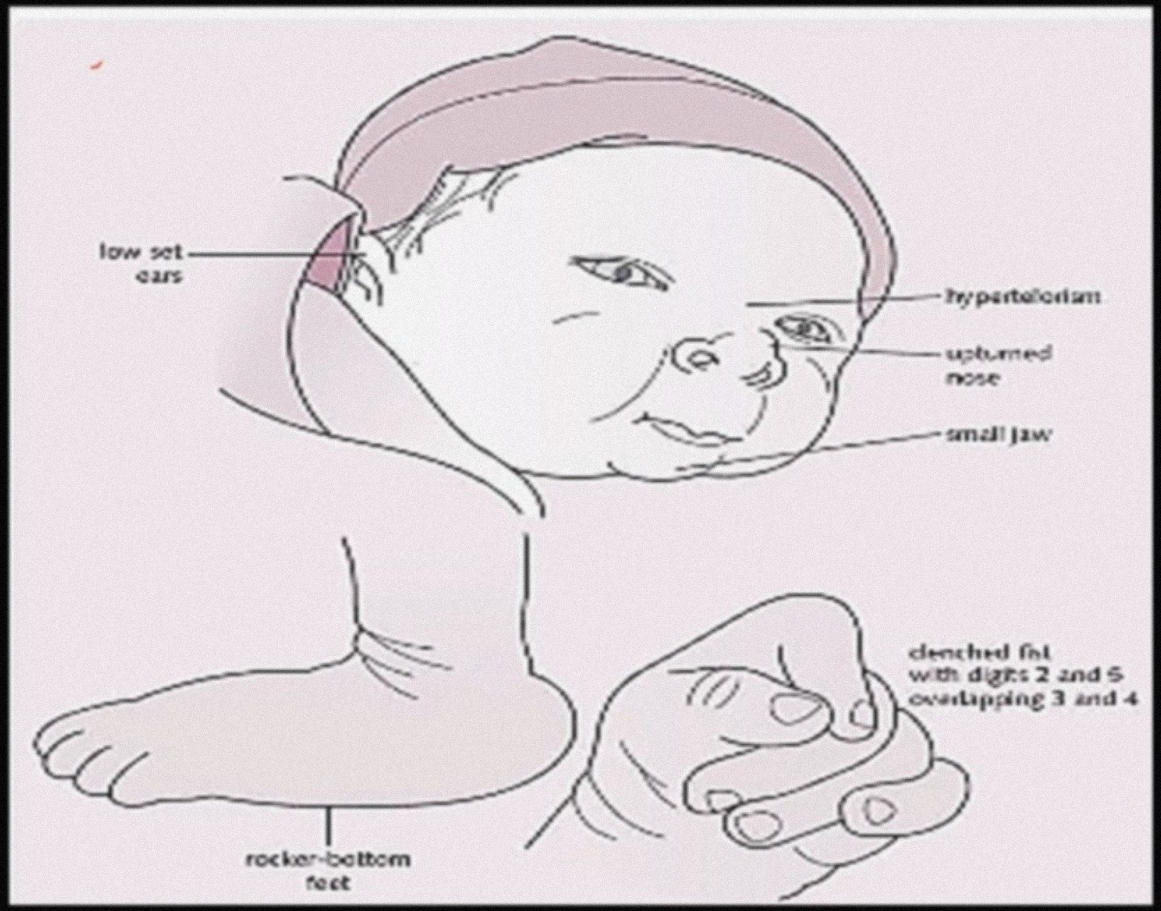
- Cleft lip
- Cleft palate
- Hypotelorism
- Low set ears
- Post axial polydactyly
- Microphthalmia
- Heart defects



TRISOMY 18 / EDWARDS SYNDROME

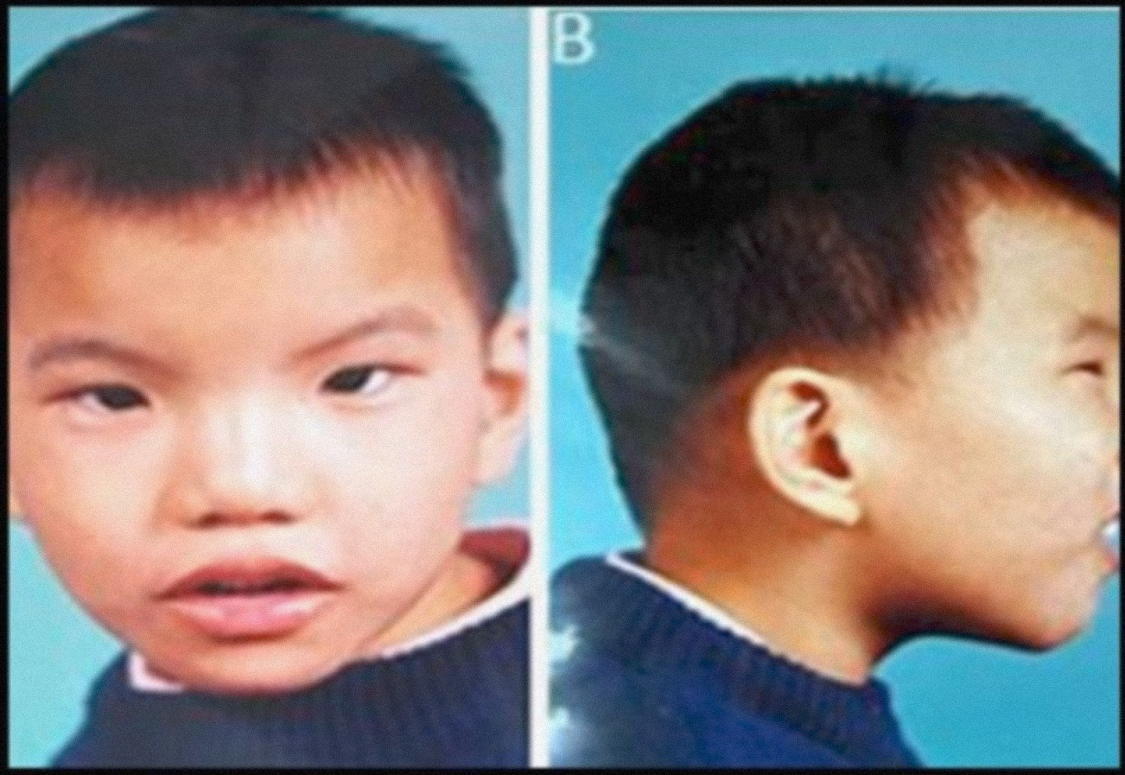
- Cataract include ptosis
- Microphthalmos
- Corneal opacity
- **Uveal and disc coloboma**
- Vitreoretinal dysplasia

- Rocker bottom feet
- Clenched fist with 2nd and 5th overlapping 3rd and 4th

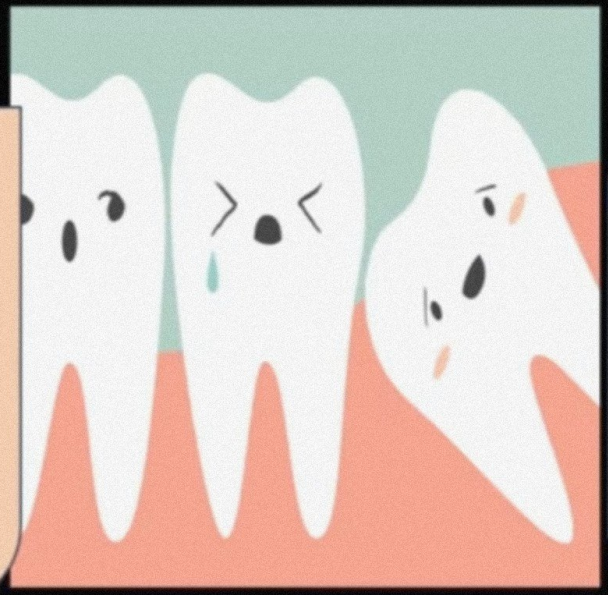


CRI-DU-CHAT SYNDROME

- Downward slant
- Widely set eyes (hypertelorism)
- Low-set ears,
- Small jaw, and a rounded face.

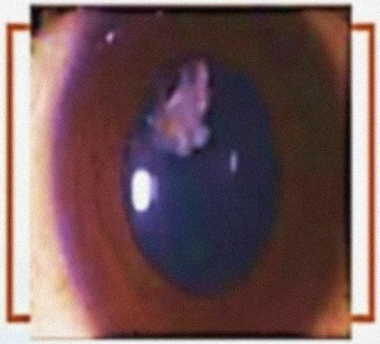


NANS HORAN SYNDROME

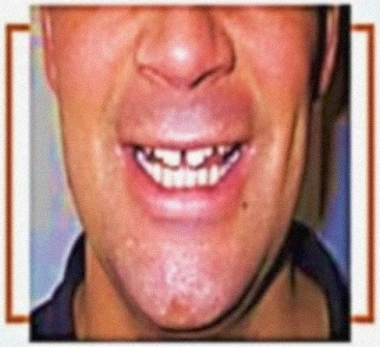


LENS MICROPHTHALMIA SYNDROME

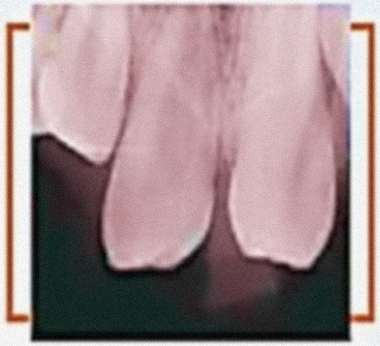
NANS HORAN SYNDROME



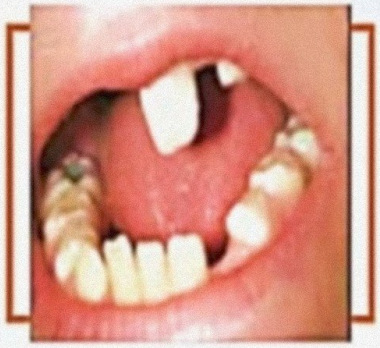
Microcornea, micropthalmia



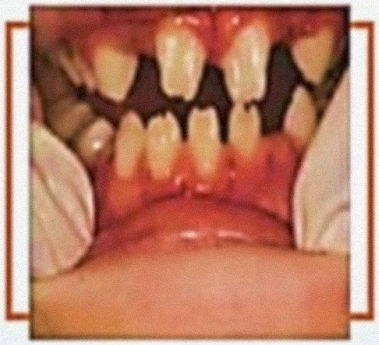
Small teeth in males



Canines: dome shaped with trilobed edge.



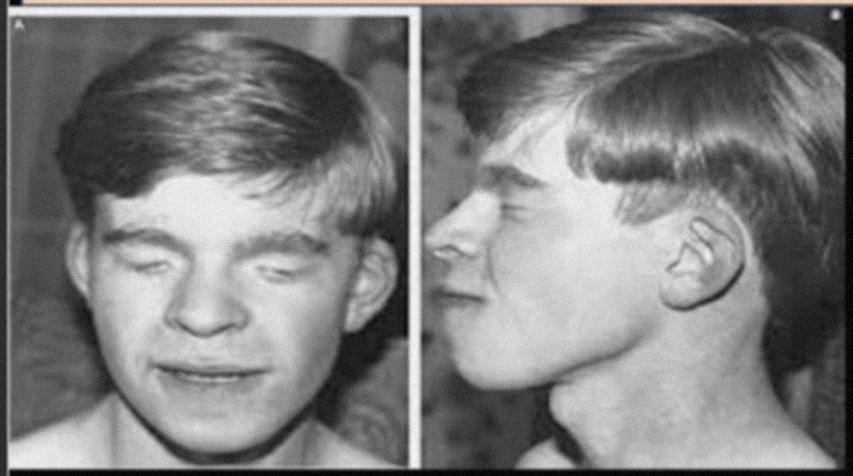
Premolars & molars: small, round, mulberry shaped.



Incisors: screwdriver shaped/conical, irregular incisal edge

Y SHAPED SUTURE CATARACT

NANS HORAN SYNDROME



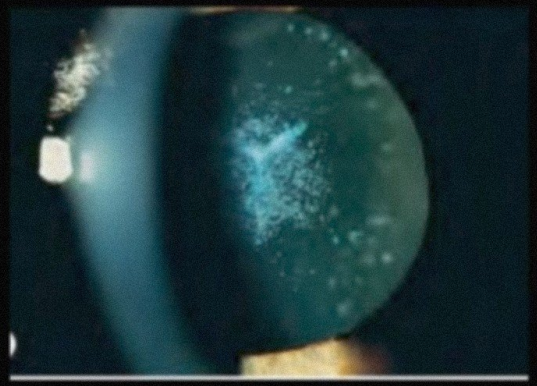
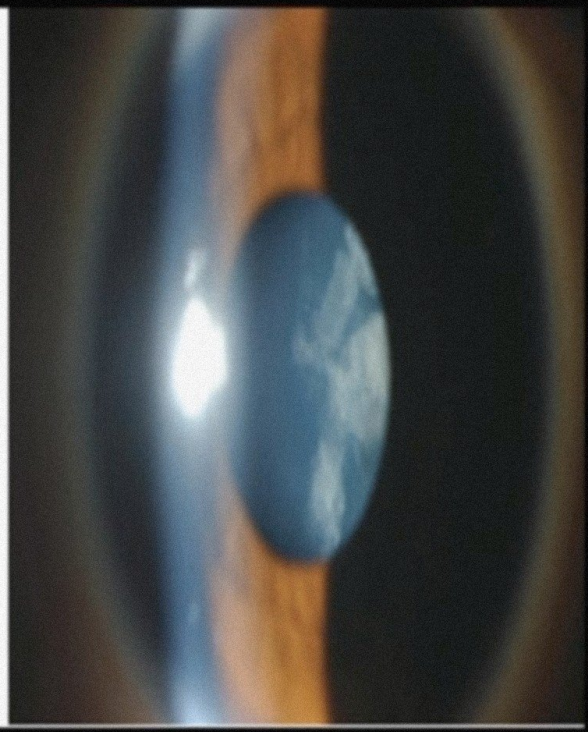


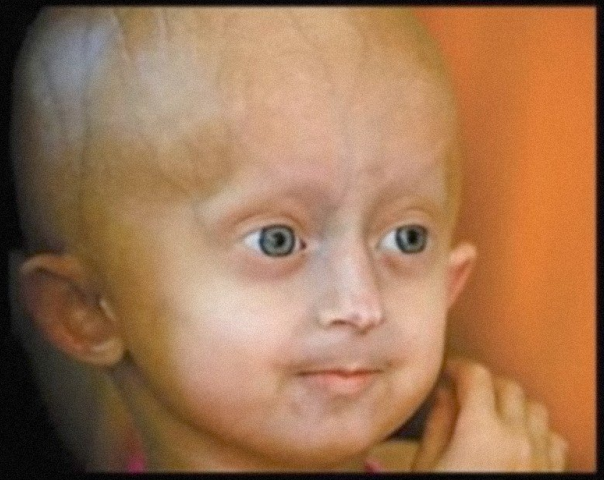
Figure 22-1 The asymptomatic sister of a boy with suspected Nance-Horan syndrome was examined for potential female carrier signs. Both of her eyes showed partial sutural lens opacity (left eye shown). Genetic testing confirmed the boy's diagnosis and his sister's carrier state. *(Courtesy of Arif O. Khan, MD.)*

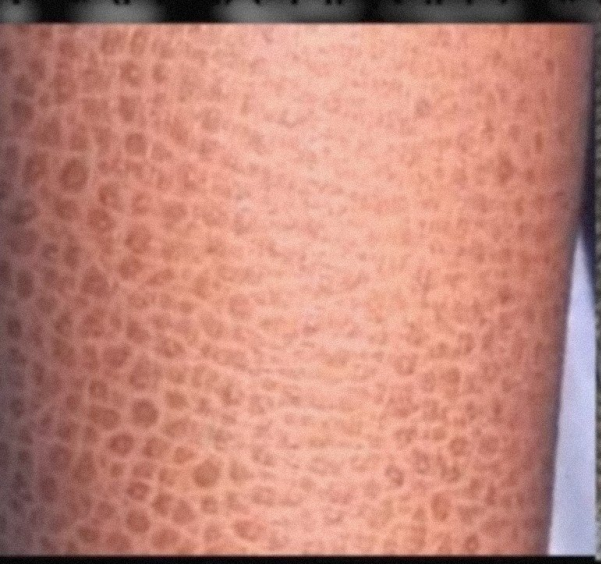
SOURCE : AAO



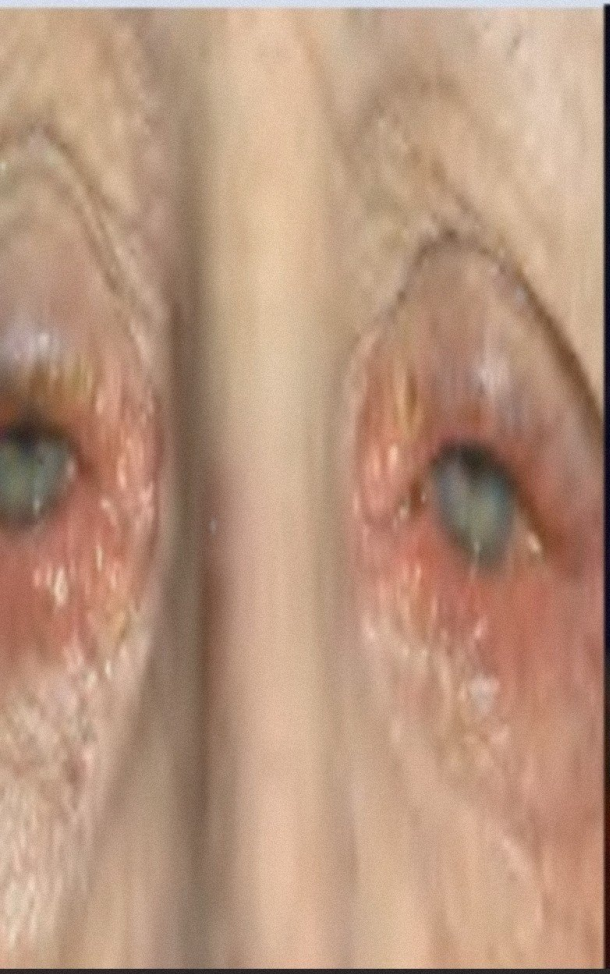
SKIN DISORDERS with Cataracts

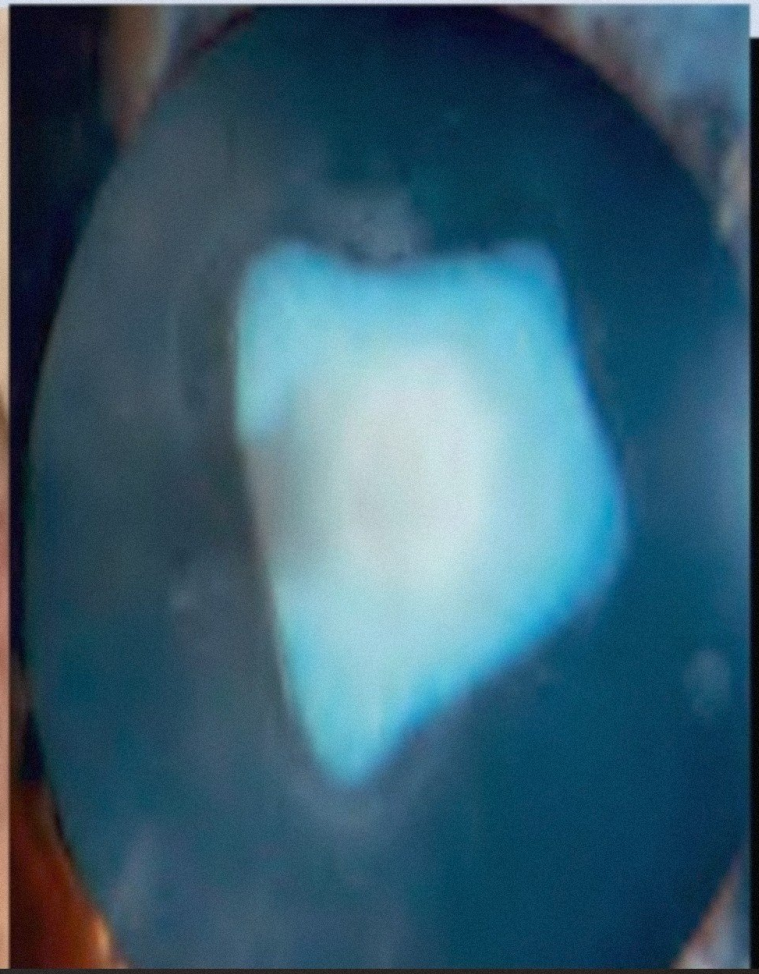
- Cockayne syndrome
- Rothmund-Thomson
- Atopic dermatitis
- Incontinentia pigmenti
- Progeria
- Ichthyosis





ATOPIC DERMATITIS CATARACT





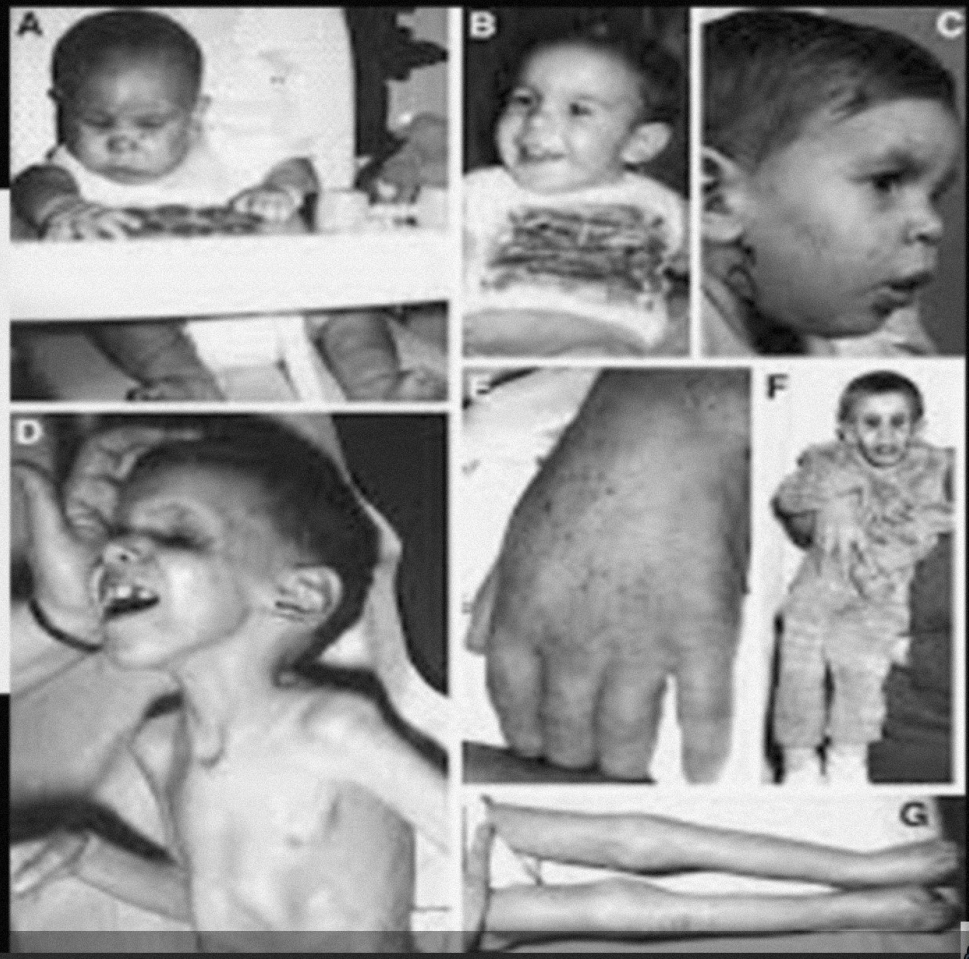
- About 10%
- second to fourth decades.
- Bilateral and may mature quickly.

Shield-like dense anterior subcapsular plaque that wrinkles the anterior capsule is characteristic.

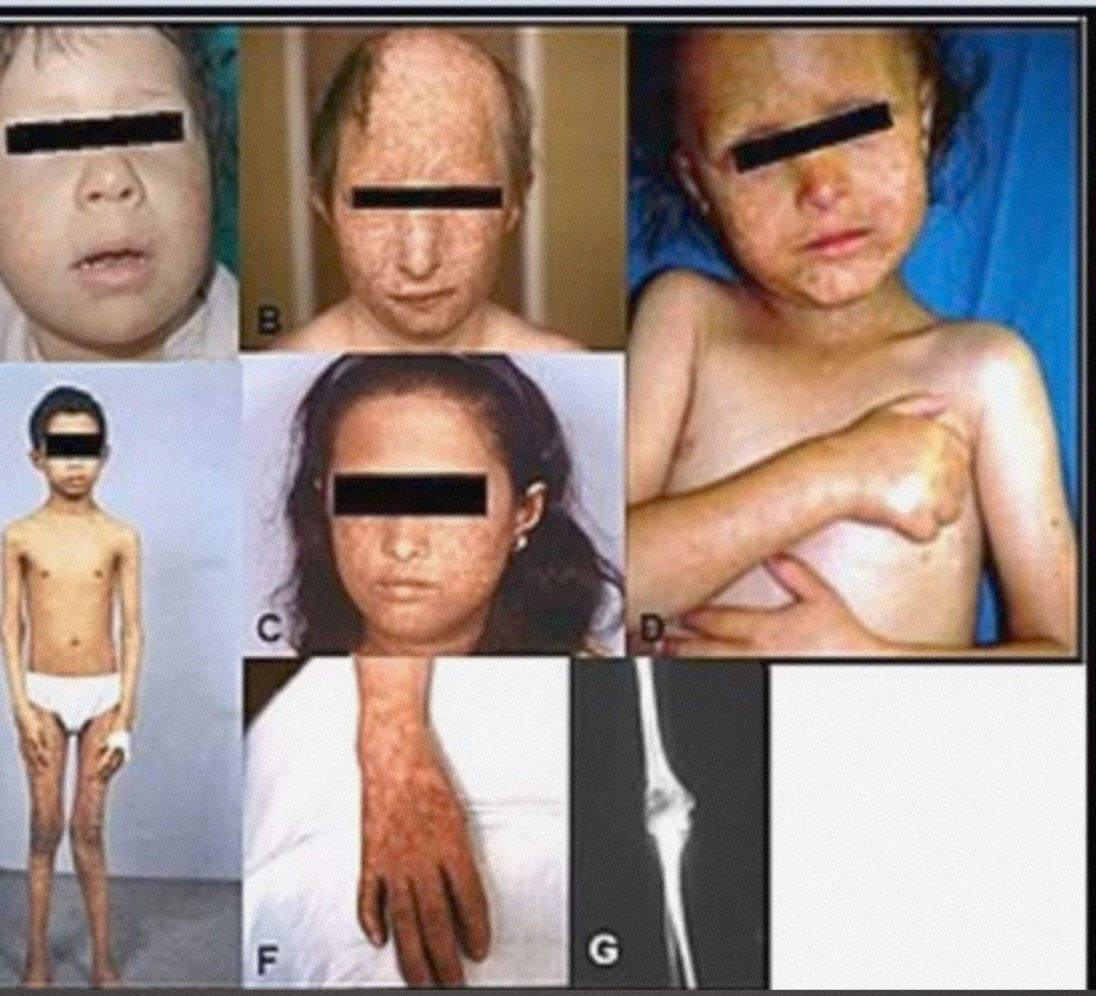
- Posterior subcapsular opacities may also occur

COCKAYNE SYNDROME

The skin of those with Cockayne syndrome is also frequently affected: **hyperpigmentation**, **varicose** or spider veins (**telangiectasia**), and serious sensitivity to sunlight are common, even in individuals without XP-CS

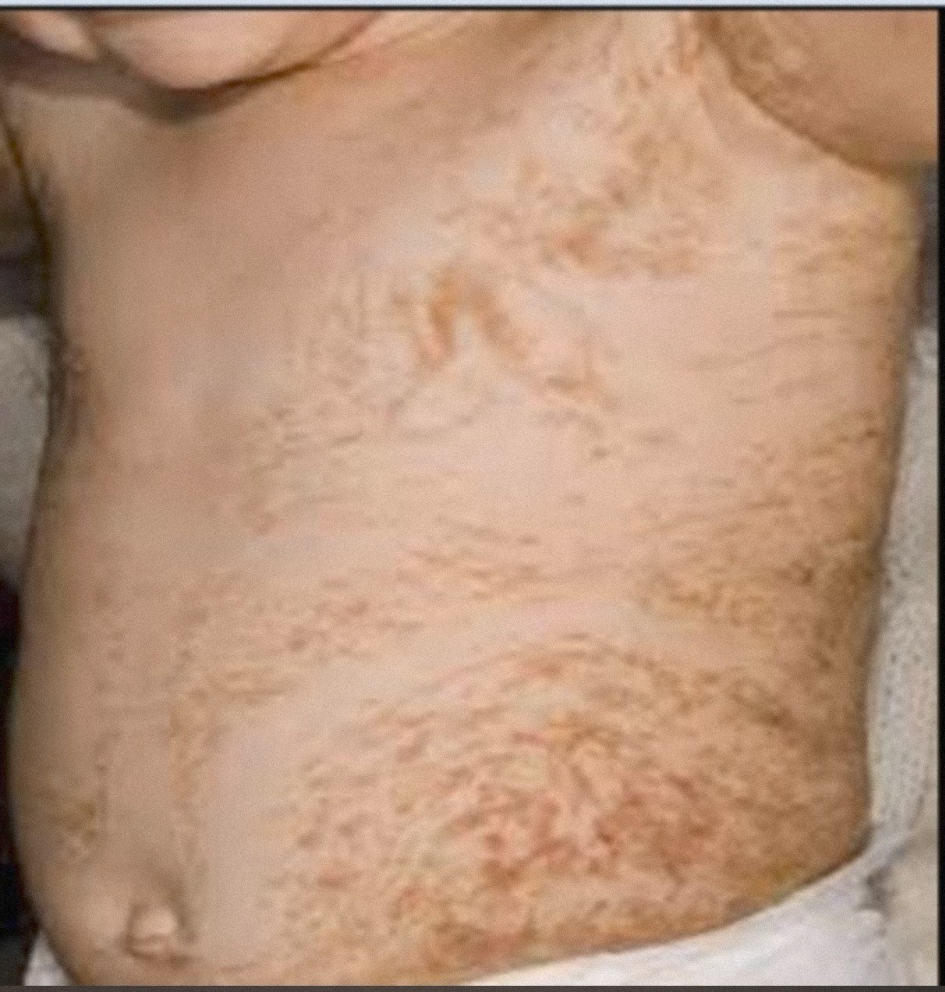


OTHMUND-THOMSON



Poikiloderma is a skin condition that consists of areas of hypopigmentation, hyperpigmentation, telangiectasias and atrophy.

INCONTINENTIA PIGMENTI



1. **BLISTERING** (from birth to about four months of age),
2. A **WART-LIKE** rash (for several months),
3. **SWIRLING** macular hyperpigmentation (from about six months of age into adulthood), followed by
4. Linear **HYPOPIGMENTATION**.