BILATERAL CATARACTS

GENETIC MUTATION

METABOLIC SYNDROMES

SYSTEMIC SYNDROMES

CHROMOSOMAL ANOMALIES

TORCH INFECTION

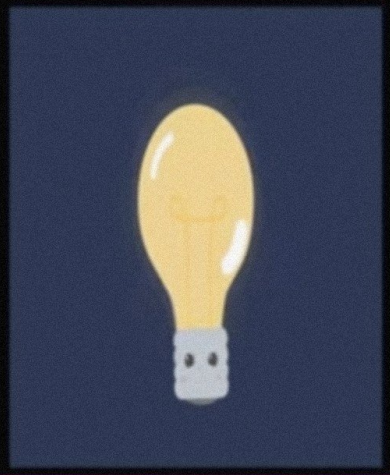
GENETIC MUTATION

- **PAX 6**
- **FOX E3**
- **C MAX**
- **PITX 3**
- **MIP**

Genes related to connexins and crystallins of the lens

Inherited without systemic abnormalities

- **AUTOSOMAL DOMINANT** (most common)
- **AUTOSOMAL RECESSIVE** (mostly seen in families with a history of consanguinity)
- **X-LINKED**

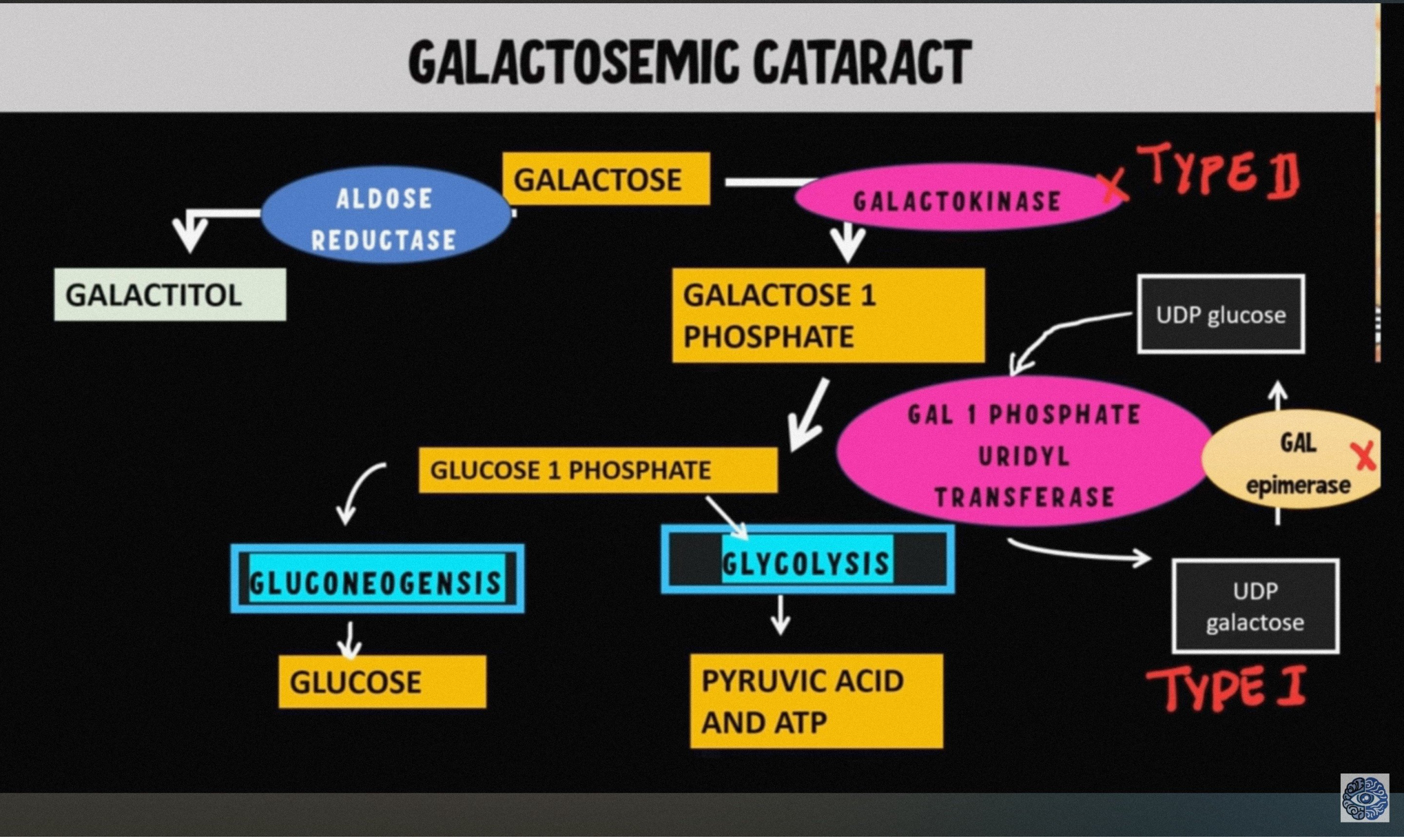


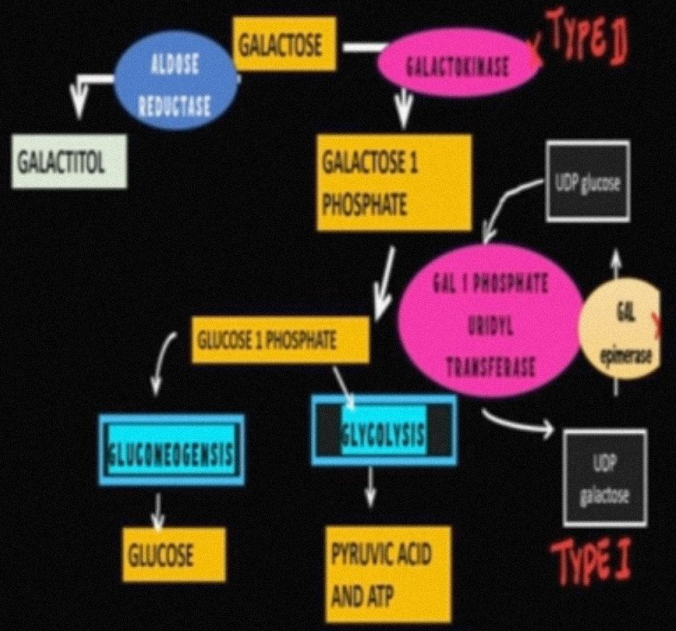
Isolated inherited congenital cataracts carry a better visual prognosis than those with coexisting ocular and systemic abnormality

GALACTOSEMIC CATARACT

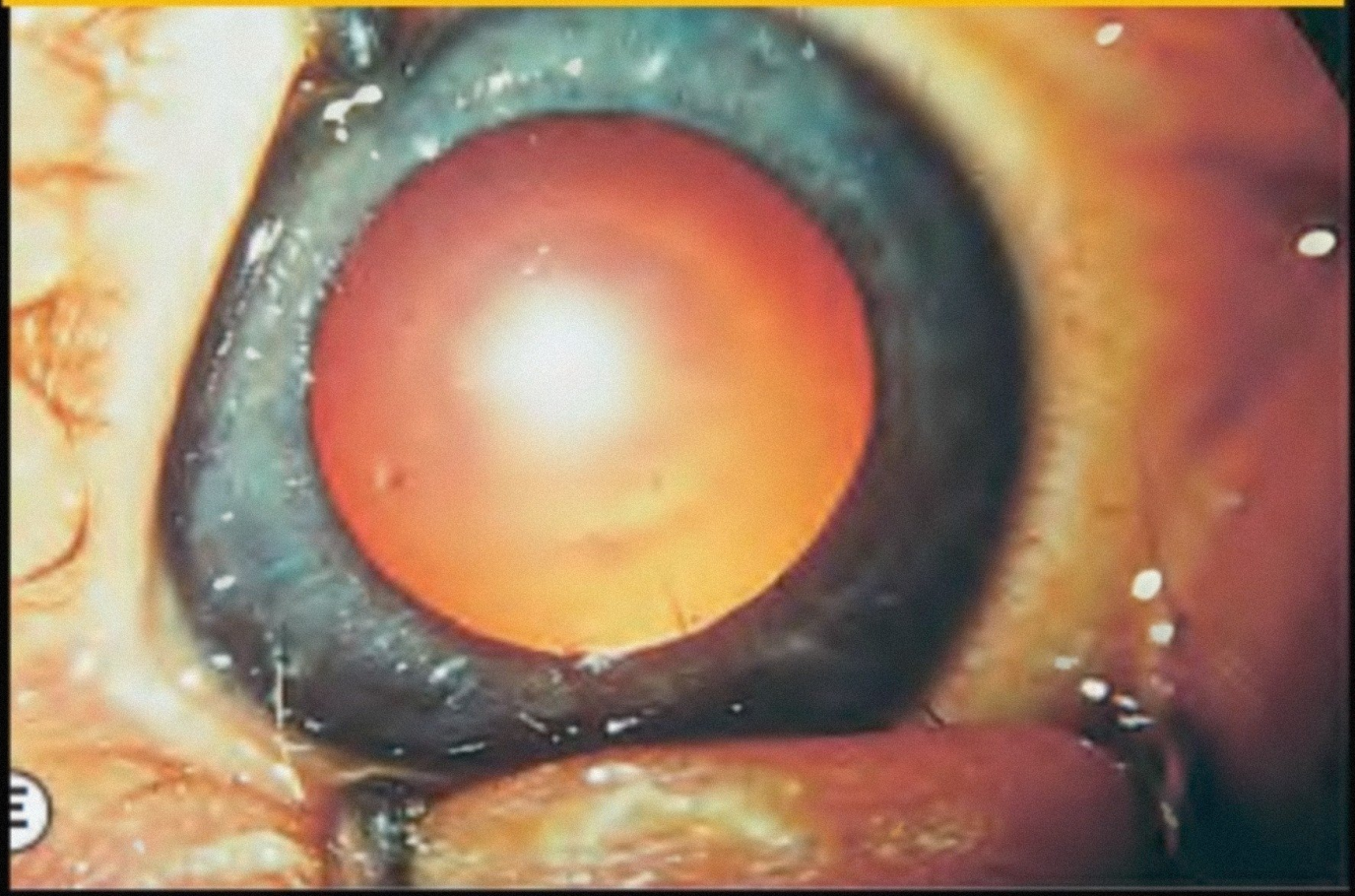
This is an **AUTOSOMAL RECESSIVE**, inherited congenital disease characterized by an inborn inability of the infant to metabolize galactose

GALACTOSEMIC CATARACT





OIL DROP CATARACT



METABOLIC CATARACT

GALACTOSEMIA

FABRY DISEASE

DIABETIC CATARACT

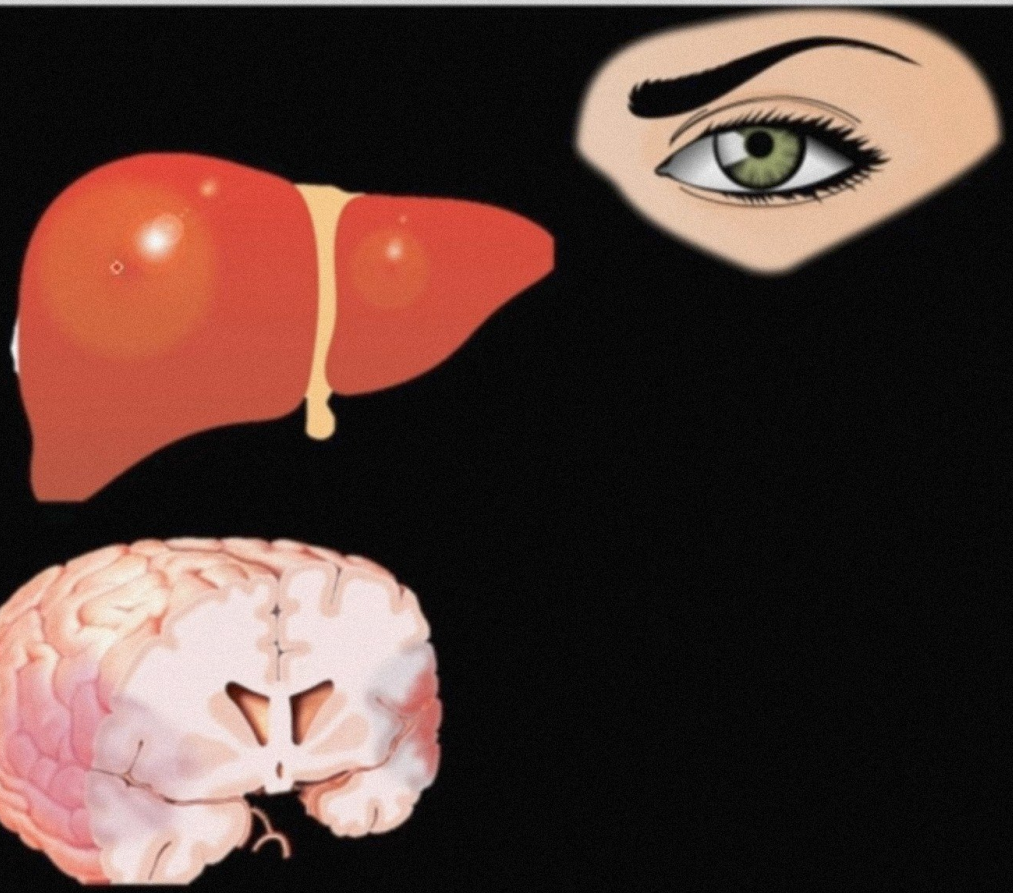
WILSON DISEASE

LOWE DISEASE

HYPOCALCEMIC / PARATHYROID TETANY RELATED

WILSON DISEASE

- **HEPATOLENTICULAR DEGENERATION**
- Inherited disorder of **copper** metabolism due
- Mutation of the **ATP7B** gene.
- Affects eye, liver and basal ganglion.



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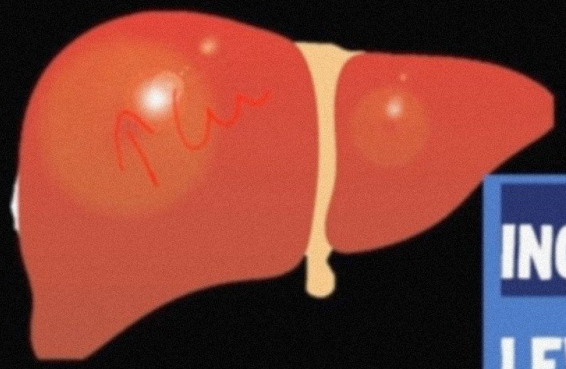
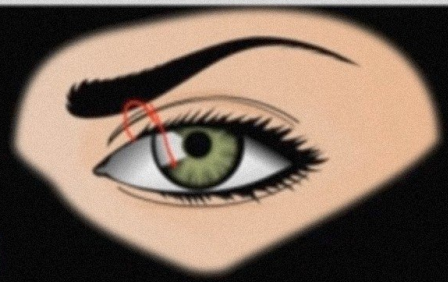
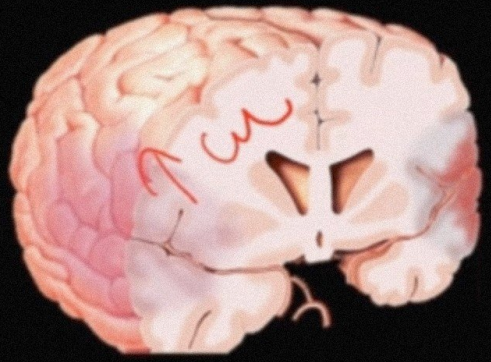
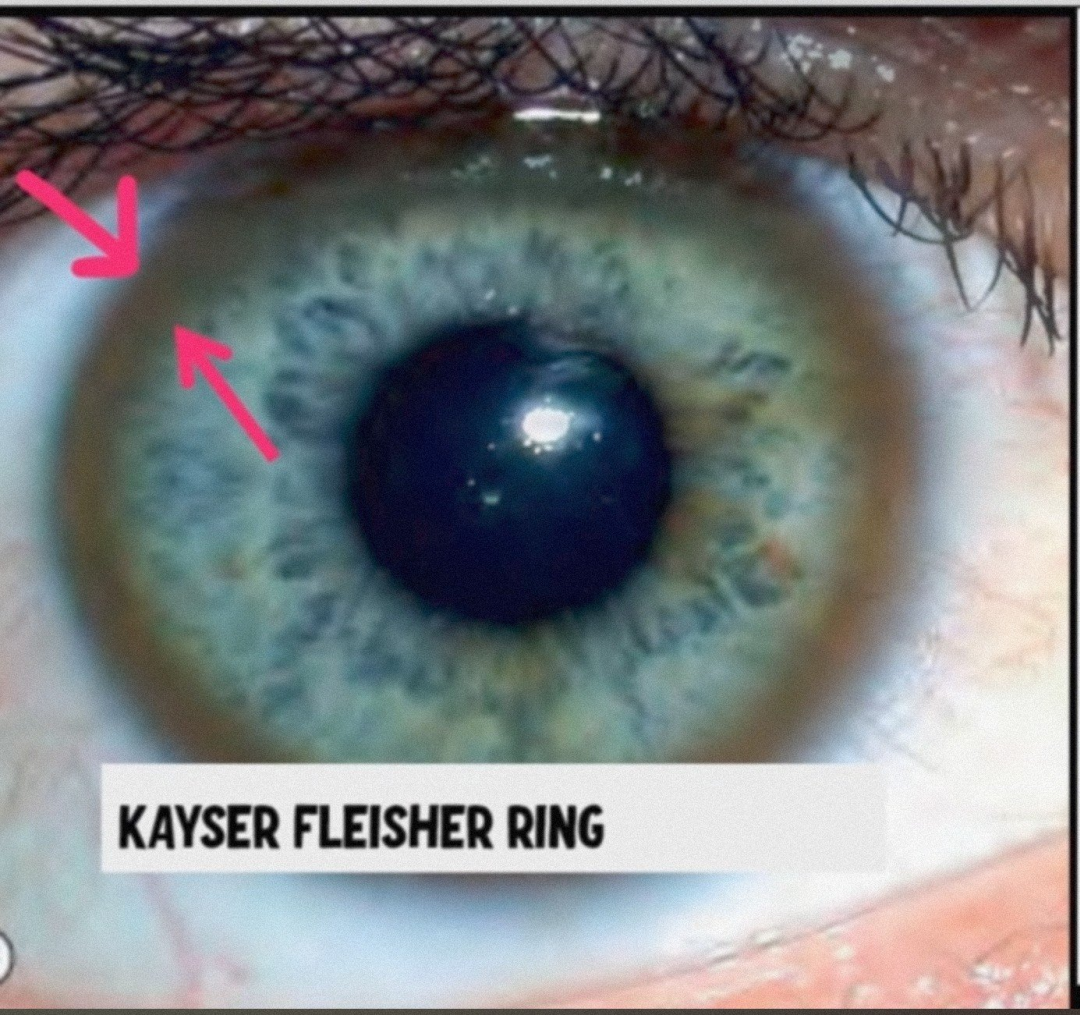






INCREASED COPPER LEVELS AND DECREASED CERULOPLASMIN LEVELS

WILSON DISEASE



KAYSER FLEISHER RING

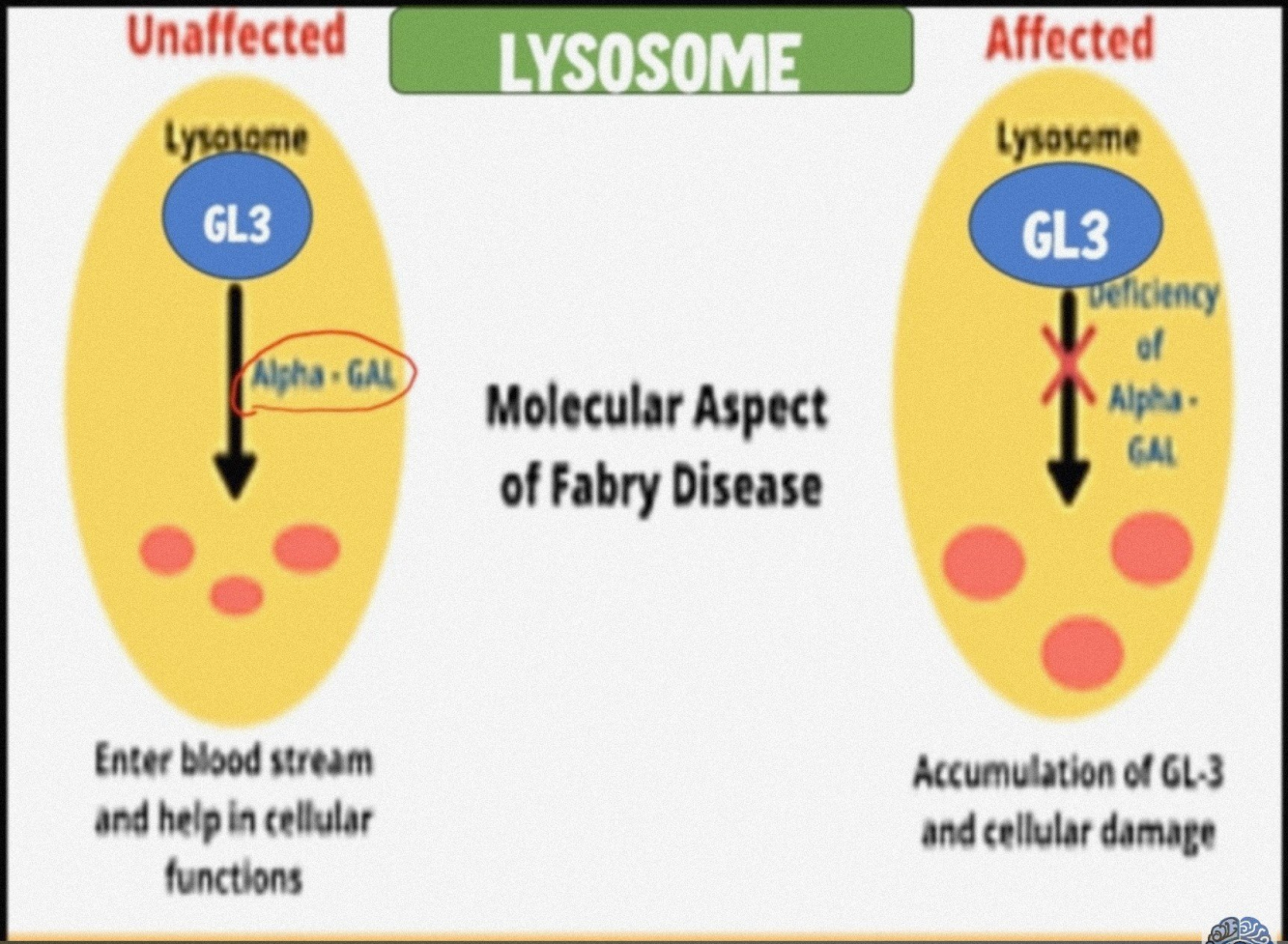
SUNFLOWER CATARACT

• FIG. 28.16 An intraocular copper foreign body causing a sunflow cataract. Foreign bodies that cause chalcosis are usually compose of less than 85% copper. Pure copper causes a suppurative endophthalmitis. Source: (From Jay H. Krachmer, David A. Palay eds. *Cornea Atlas*, 3rd ed. London: Saunders; 2014. pp. 243–271.

FABRY DISEASE

- **X-linked lysosomal storage** disorder
- Deficiency of the enzyme **alpha-galactosidase A**
- This leads to abnormal tissue accumulation of a **glycolipid**

All males with the gene develop the disease and some heterozygous females

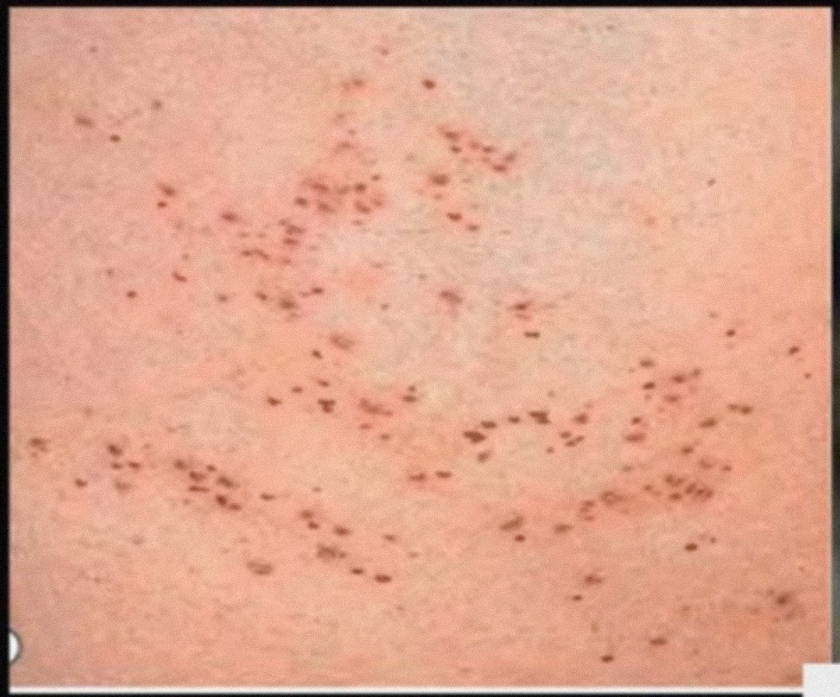


Fabry's Disease:

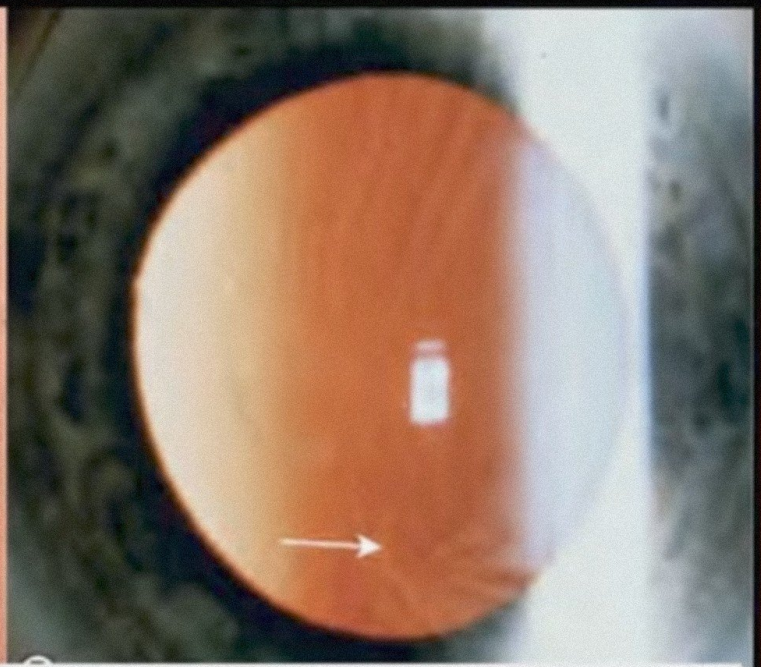
FABRY'C (*replace S with C*)

- **F**oam cells / **F**ebrile episodes
- **A**lpha galactosidase **A** deficiency / **A**ngiokeratomas
- **B**urning pain in hands & feet "Peripheral neuropathy" / **B**oys
- **R**enal Failure
- **Y**X genotype (Male, X-linked recessive)
- **C**eramide trihexoside *accumulation* / **C**ardiovascular disease

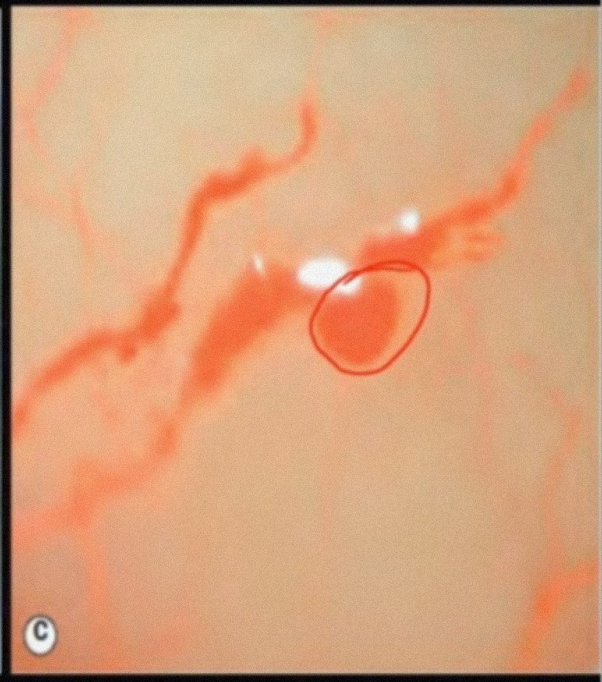
FABRY DISEASE



ANGIOKERATOMAS



VERTICELLATA / Vortex keratopathy

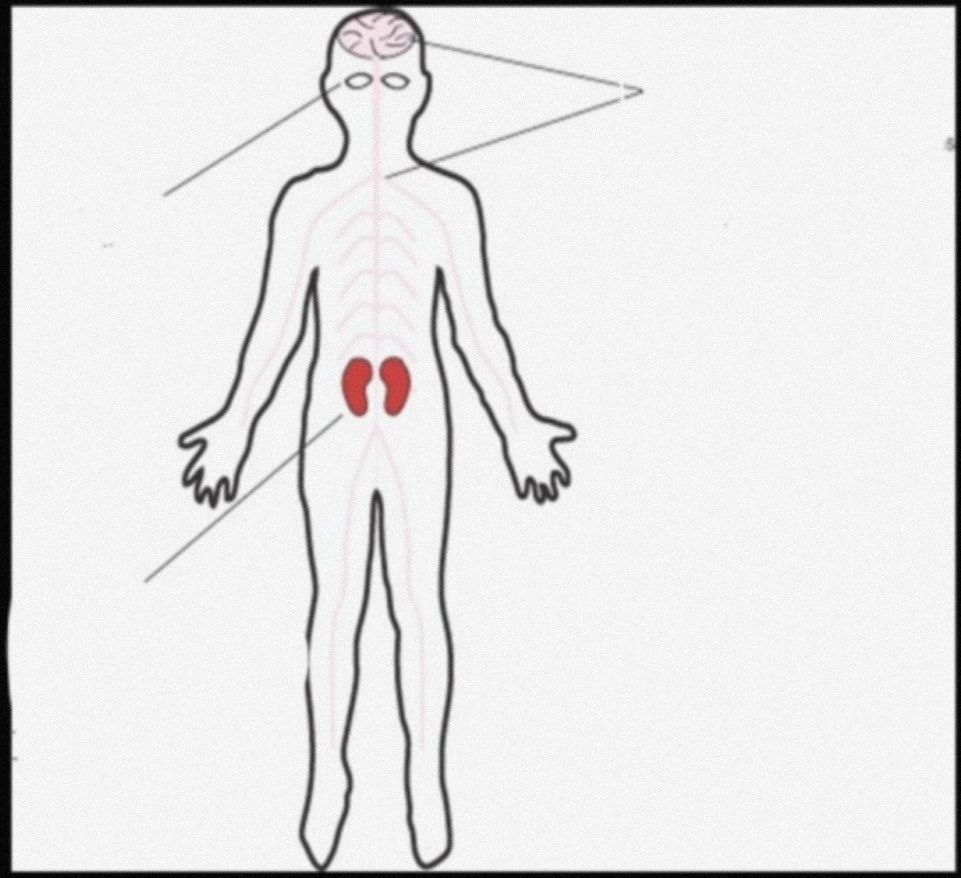


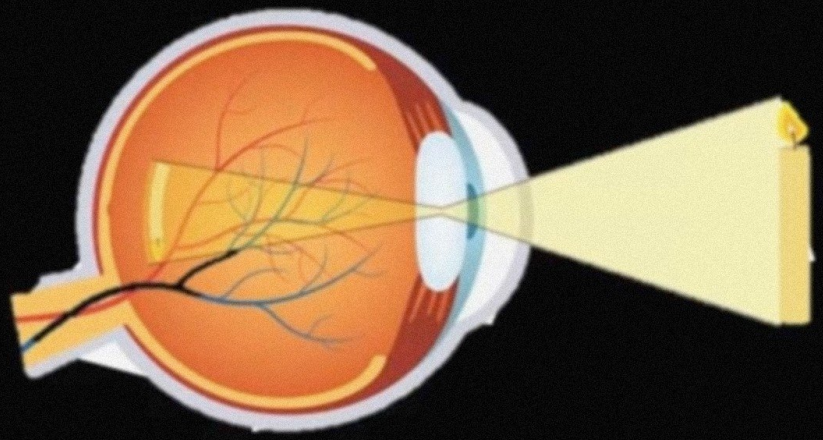
CORKSCREW VESSELS

LOWE DISEASE

- Lowe syndrome (**oculocerebrorenal**) syndrome
- **X-linked** recessive (gene: OCRL1) inborn error of amino acid metabolism
- Neuromuscular, renal and other manifestations.

POSTERIOR LENTICONUS



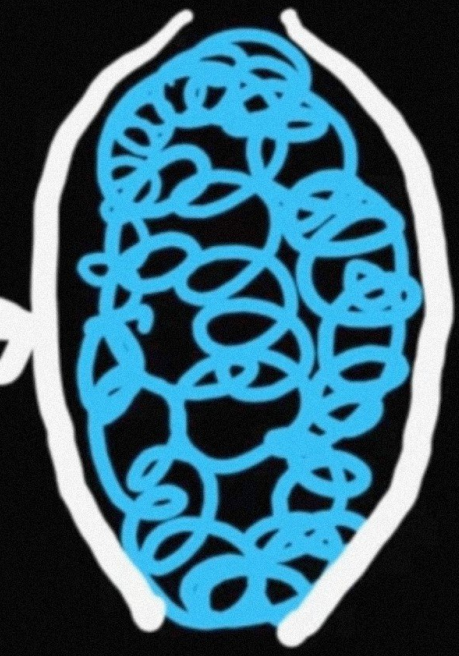


CHANGE OF REFRACTION →

MYOPIA

SORBITOL

HYDRATED LENS



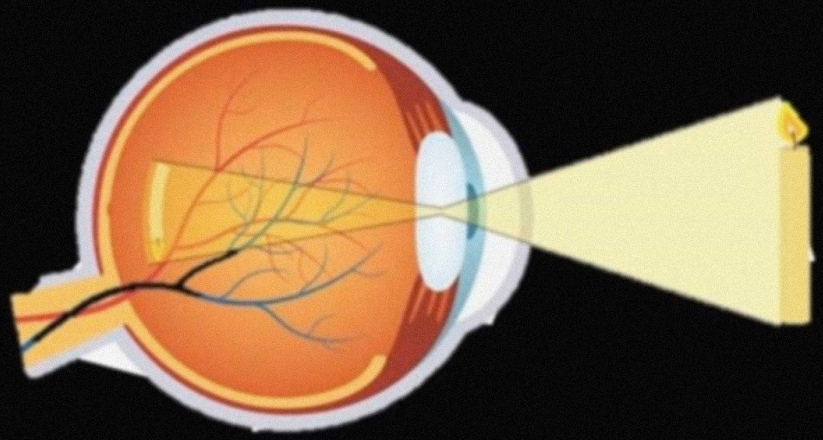
TRUE DIABETIC CATARACT

GLUCOSE → ALDOSE REDUCTASE

SORBITOL: retained within the lens → increased OSMOTIC GRADIENT

Water entry \ Hydration of the lens → Vacuole formation and swelling

OPACIFICATION AND CATARACT



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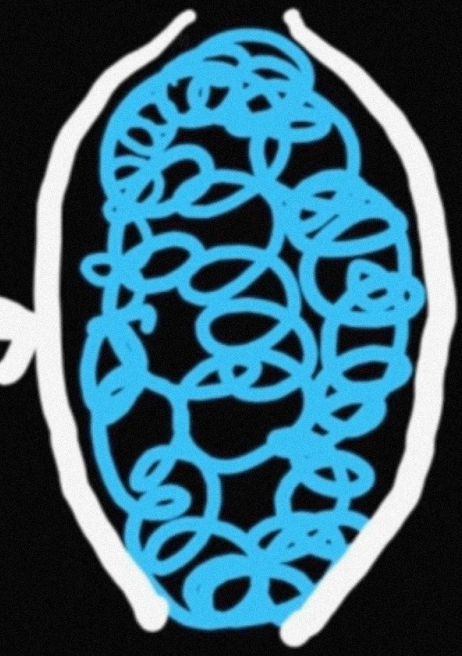

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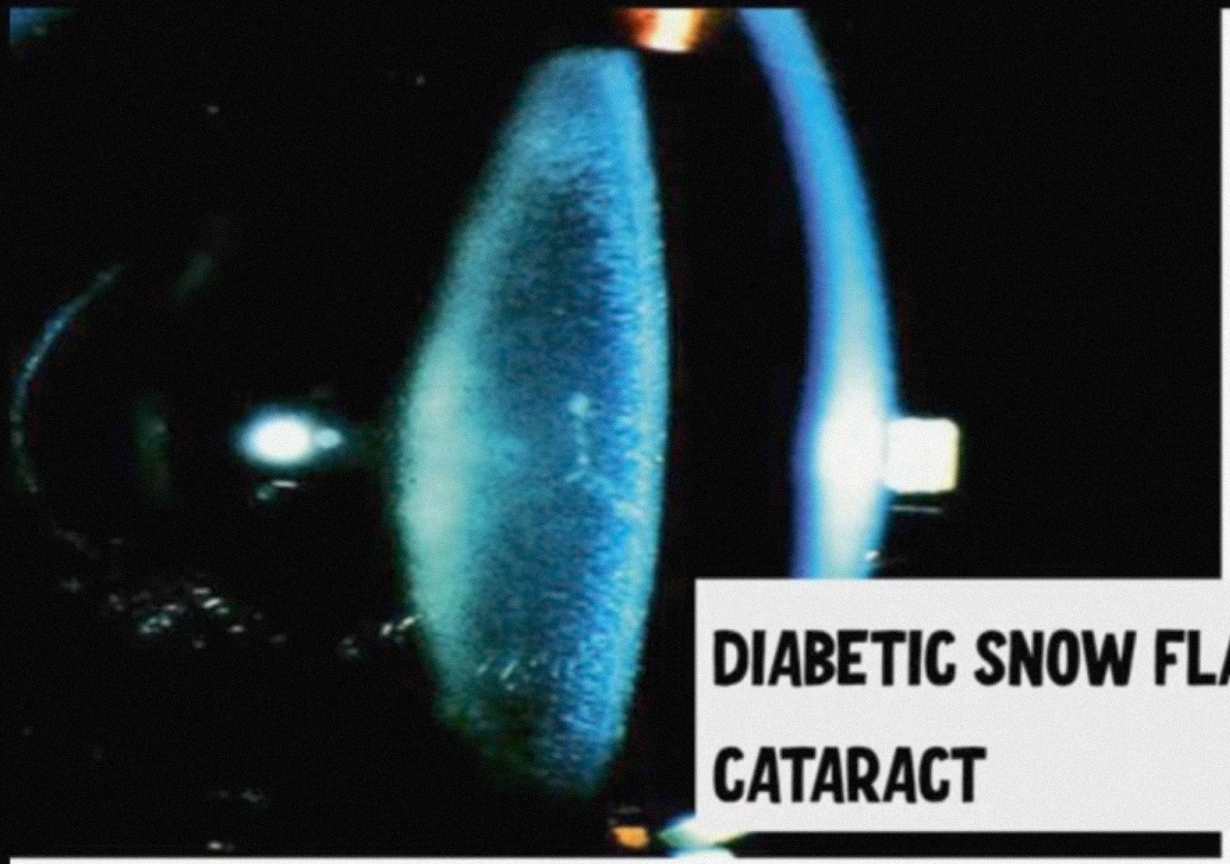
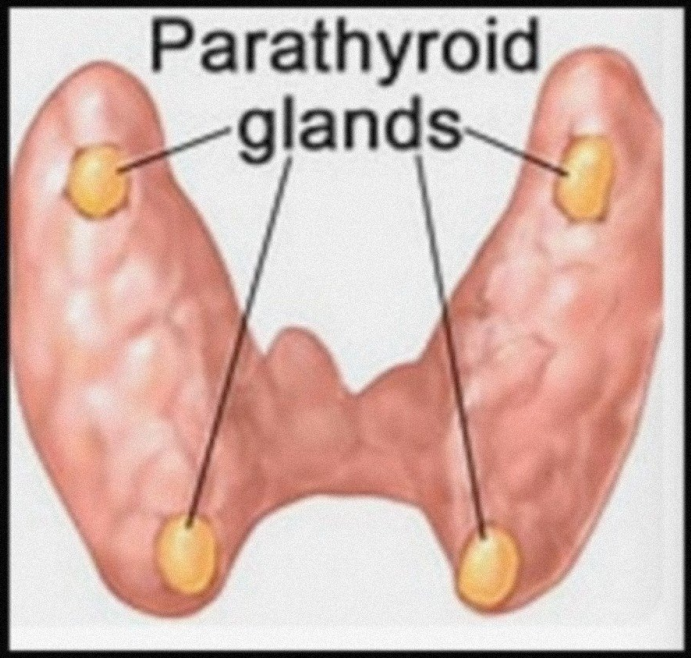


DIABETIC SNOW FLAKE CATARACT

G. 15.7 Cataract in systemic disease. Diabetic snowflake
ct. (*Source:* From Jack J. Kanski, Brad Bowling, eds. *Clinical*
lmology: A Systematic Approach, 7th ed. London: Saunders;

HYPOCALCEMIA / PARATHYROID TETANY



- Atrophy
- Removal of parathyroids
- Hypocalcemia

This affects the membrane of the lens basically .

An **sodium concentration** which is maintained low also increases in the lens leading to the cataract