FABRY DISEASE
SECOND STAGE

DR. RASHAD AL - TUUAMAH
MEDICAL BIOCHEMISTRY

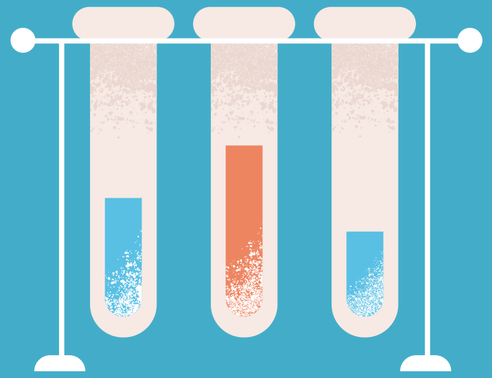
Introduction and Definition

FABRY disease is a rare X-linked lysosomal storage disorder caused by deficiency of the enzyme ALPHA-GALACTOSIDASE A.

this leads to accumulation of GLOBOTRIAOSYLCERAMIDE (GB3) in cells.

it affects multiple organs: kidneys, heart, nervous system, skin, and sometimes oral tissues.

FABRY disease is a rare inherited disorder causing progressive multi-organ damage.



GLA Gene - Name, Sequence and Details

the gene name is GLA (galactosidase alpha) located at chromosomal location Xq22.1.

its NCBI GENE ID is 2717. the gene length is approximately 14,740 bases with a coding region of 1290 bp across 7 exons. the protein

encoded is ALPHA-GALACTOSIDASE A (429 amino acids). imagine the X chromosome as a huge book; the GLA gene is like a special line

in this book at Xq22.1. the important lines, called coding regions (exons), turn into the ALPHA-GALACTOSIDASE A enzyme which

acts to clean up fat. mutations in the GLA gene cause GB3 accumulation and FABRY disease.

Prevalence and Global Statistics
the prevalence (current cases) shows it is a rare disorder affecting 1 in 40,000 – 1

in 117,000 people worldwide. many cases are likely underdiagnosed. in IRAQ, there are 17 confirmed cases among 1,148 screened.

FABRY disease is rare globally and in IRAQ, but true numbers may be higher due to underdiagnosis.

Incidence and Newborn Screening

the incidence (new cases per year) estimated from newborn screening is 1 in 3,000 - 7,800 male births.

these numbers vary by country and detection methods. early diagnosis is crucial for management and treatment. newborn screening

studies show more cases, emphasizing the need for awareness and early genetic testing.

Dental Manifestations and Oral Symptoms

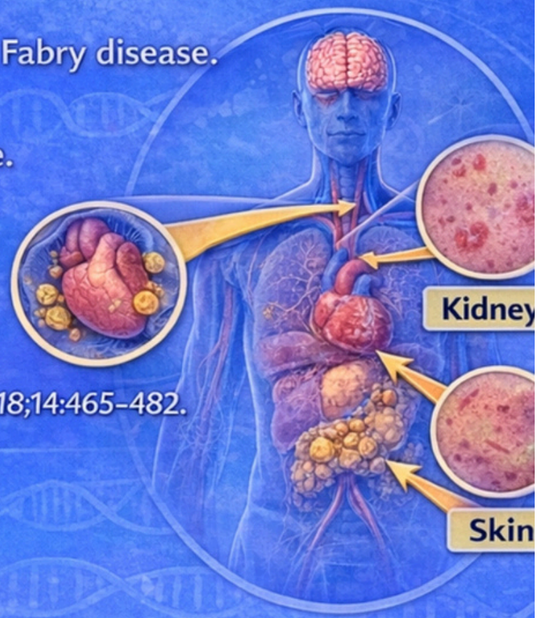
oral symptoms in FABRY disease include burning mouth sensation, reduced salivary flow (XEROSTOMIA), and difficulty chewing and swallowing due to neuropathic pain. dental symptoms also include increased dental caries risk, gingival inflammation, and ANGIOKERATOMAS on gums.

Importance of Early Diagnosis

early diagnosis is essential as it prevents irreversible organ damage, improves quality of life, and allows timely treatment for better long-term outcomes.

References

- **Review Articles:**
  - Germain DP. Fabry disease. *Orphanet J Rare Dis*. 2010:5:30.
  - Desnick RJ, Ioannou YA, Eng CM. *α-Galactosidase A deficiency*: Fabry disease. *Metab Mol Bases of Inher Dis*. 2001;8th ed.
  - Kampmann C, Linhart A. Car*diac manifestations* of Fabry disease. *Heart*. 2015;101:1–7.
  - MacDermot KD, Holmes A, Miners AH. Natural history of Fabry disease in males. *J Med Genet*. 2001;38:750–760.
  - Ortiz A, Germain DP, Desnick RJ, et al. Fabry disease revisited: Management and treatment recommendations. *Nat Rev Nephrol*. 2018;14:465–482.
- **Genetic Databases:**
  - NCBI Gene – GLA: https://www.ncbi.nlm.nih.gov/gene/2717
  - OMIM – Fabry Disease: https://www.omim.org/entry/301500
- **Reference Books:**
  - Scriver CR, Beaudet AL, Sly WS, Valle D. The Metabolic & Molecular Bases of Inherited Disease. 8th ed. 2001.



Thank You for your attention !