

The path from detection to personalized long-term care for Fabry disease

Practice aid for the management of people living with Fabry disease

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Importance of early recognition of Fabry disease and its multisystem manifestations

~Age of onset

Timeline of manifestations in hemizygous male patients^{1,2}





Angiokeratomas, hypohidrosis

Gastrointestinal symptoms

Pain, febrile crises

Proteinuria, renal failure

Cardiomyopathy

Stroke



Exercise intolerance



- Diarrhoea
- Abdominal pain



- · Exercise intolerance
- · Socioeconomic impacts, depression



Haemodialysis



- Dyspnoea
- Palpitations, angina



- Tinnitus, vertigo
- Stroke sequelae



3rd decade onwards



Clinical¹

- Age of onset
- Affected organs

Phenotypic variation in Fabry disease

More than one disease?



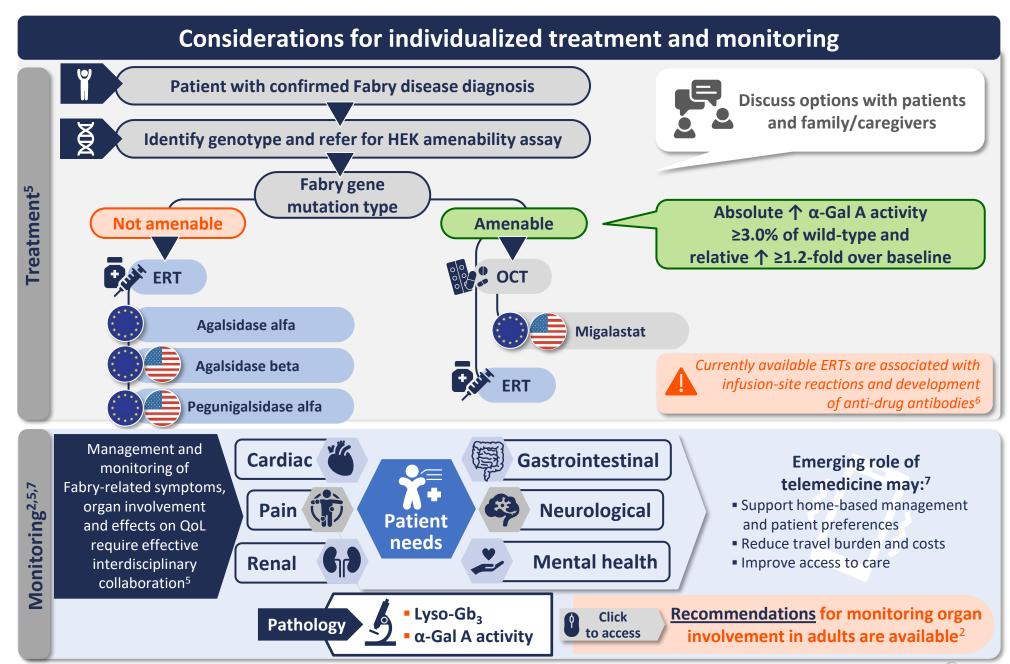
Genetic²

• Spectrum of phenotypic variants benign polymorphisms, variants of unknown significance, pathogenic variants



- Levels of lyso-Gb₃
- Residual α-Gal A activity





Abbreviations and references

Abbreviations

 α -Gal A, alpha-galactosidase A; ERT, enzyme replacement therapy; HEK, human embryo kidney; lyso-Gb₃, globotriaosylsphingosine; OCT, oral chaperone therapy; QoL, quality of life.

References

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- 2. Ortiz A, et al. *Mol Genet Metab*. 2018;123:416–27.
- 3. Arends M, et al. J Am Soc Nephrol. 2017;28:1631–41.
- 4. Lavalle L, et al. PLoS One. 2018;13:e0193550.
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- 6. Wallace EL, et al. J Med Genet. 2024;61:520–30.
- 7. Nowicki M, et al. Int J Environ Res Public Health. 2021;18:8242.

The guidance provided by this practice aid is not intended to directly influence patient care. Clinicians should always evaluate their patients' conditions and potential contraindications and review any relevant manufacturer product information or recommendations of other authorities prior to consideration of procedures, medications, or other courses of diagnosis or therapy included here.

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