

A large, stylized graphic of a globe or sphere composed of thick, orange, hand-drawn lines forming a grid pattern. The lines are slightly irregular, giving it a hand-drawn or artistic feel. It occupies the upper two-thirds of the page.

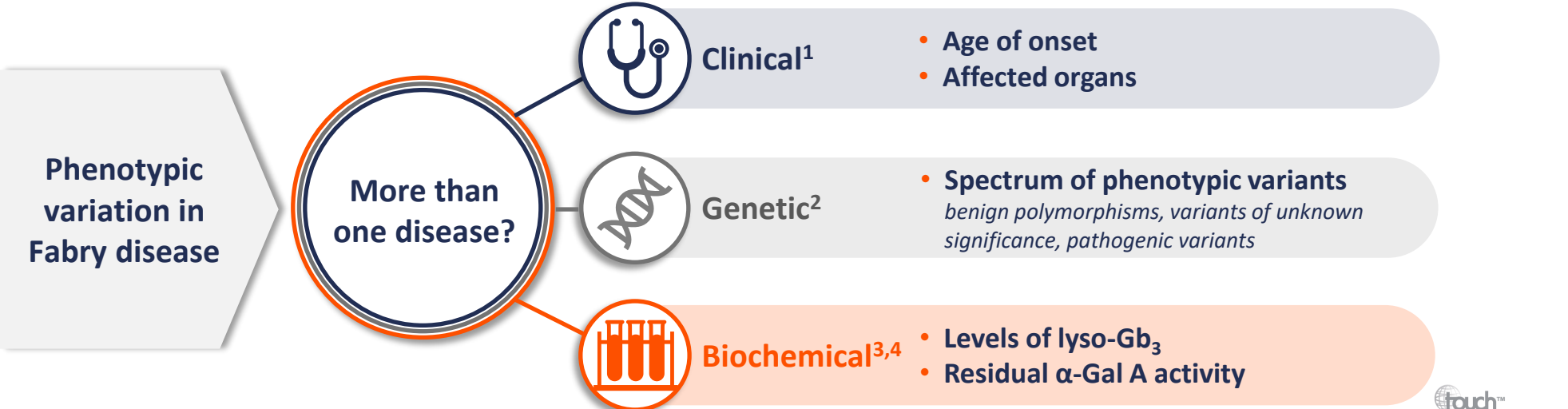
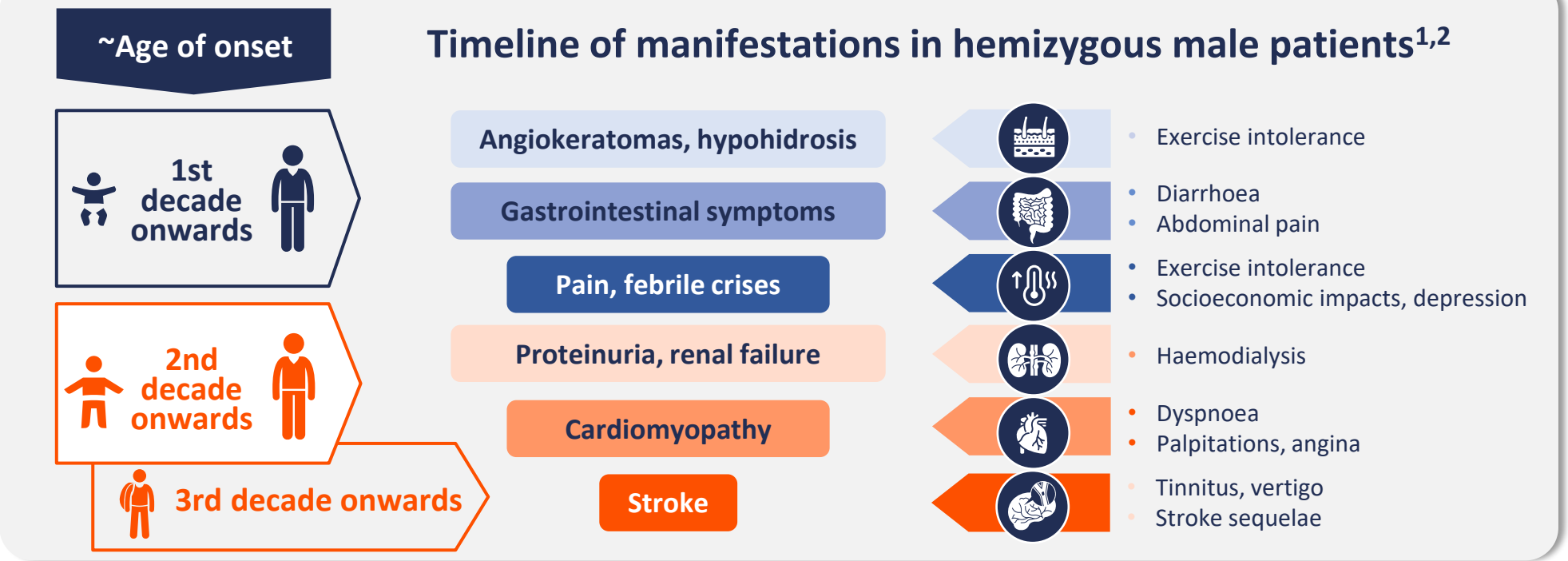
## **The path from detection to personalized long-term care for Fabry disease**

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**Practice aid for the management of people living with Fabry disease**

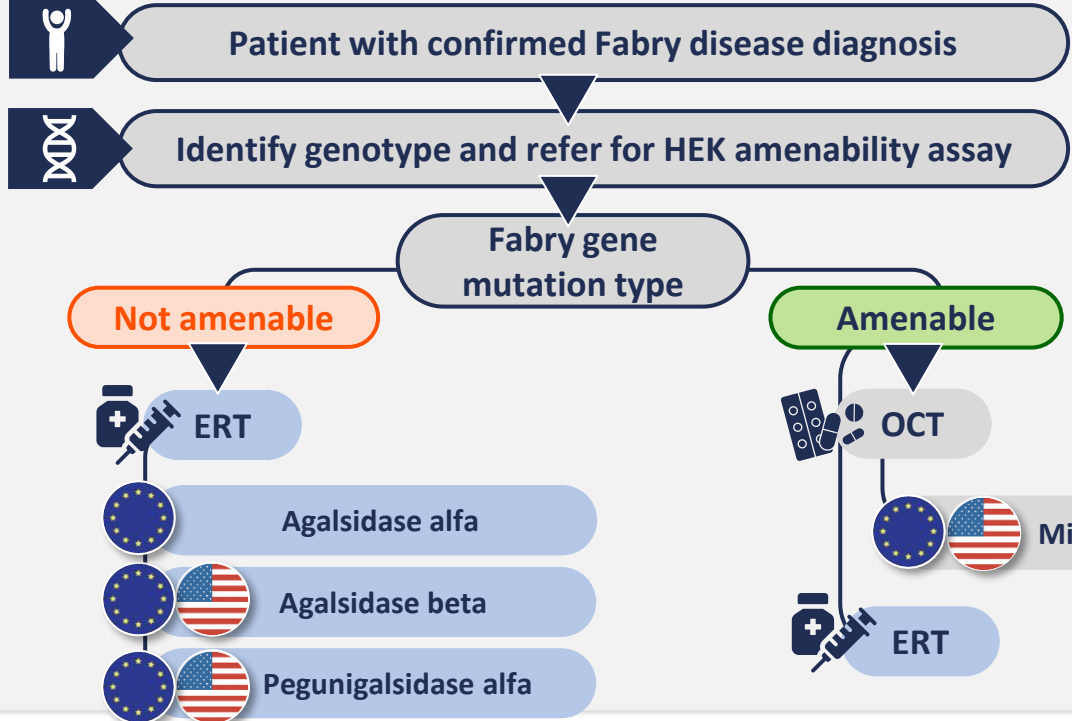
For more information, visit: [www.touchendocrinologyime.org](http://www.touchendocrinologyime.org)

# Importance of early recognition of Fabry disease and its multisystem manifestations



# Considerations for individualized treatment and monitoring

Treatment<sup>5</sup>

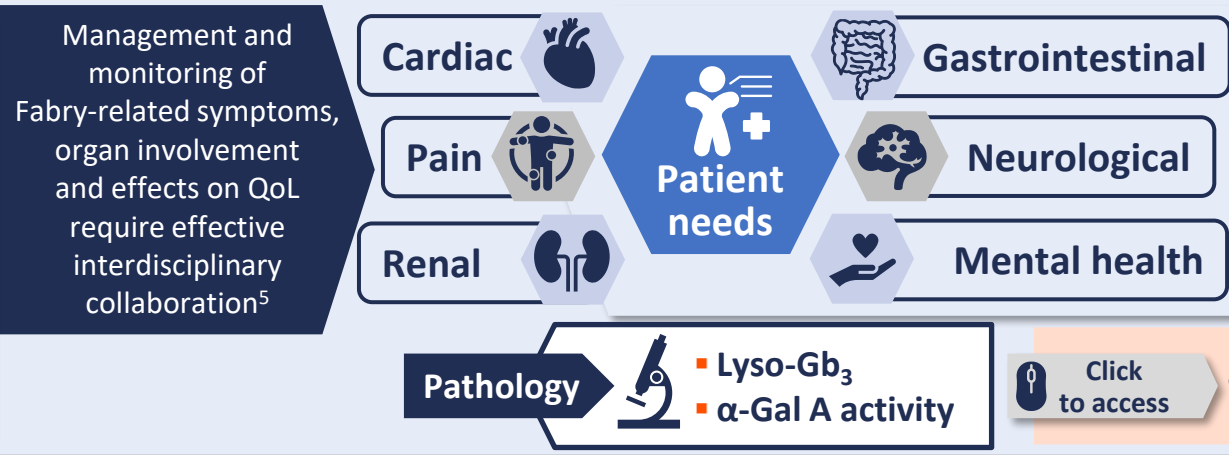


Discuss options with patients and family/caregivers

Absolute ↑ α-Gal A activity ≥3.0% of wild-type and relative ↑ ≥1.2-fold over baseline

Currently available ERTs are associated with infusion-site reactions and development of anti-drug antibodies<sup>6</sup>

Monitoring<sup>2,5,7</sup>



**Emerging role of telemedicine may:<sup>7</sup>**

- Support home-based management and patient preferences
- Reduce travel burden and costs
- Improve access to care

Click to access **Recommendations for monitoring organ involvement in adults are available<sup>2</sup>**

## Abbreviations and references

### Abbreviations

$\alpha$ -Gal A, alpha-galactosidase A; ERT, enzyme replacement therapy; HEK, human embryo kidney; lyso-Gb<sub>3</sub>, globotriaosylsphingosine; OCT, oral chaperone therapy; QoL, quality of life.

### References

1. Linhart A, Elliott PM. *Heart*. 2007;93:528–35.
2. Ortiz A, et al. *Mol Genet Metab*. 2018;123:416–27.
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4. Lavalle L, et al. *PLoS One*. 2018;13:e0193550.
5. Bichet DG, et al. *Front Med (Lausanne)*. 2023;10:1220637.
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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