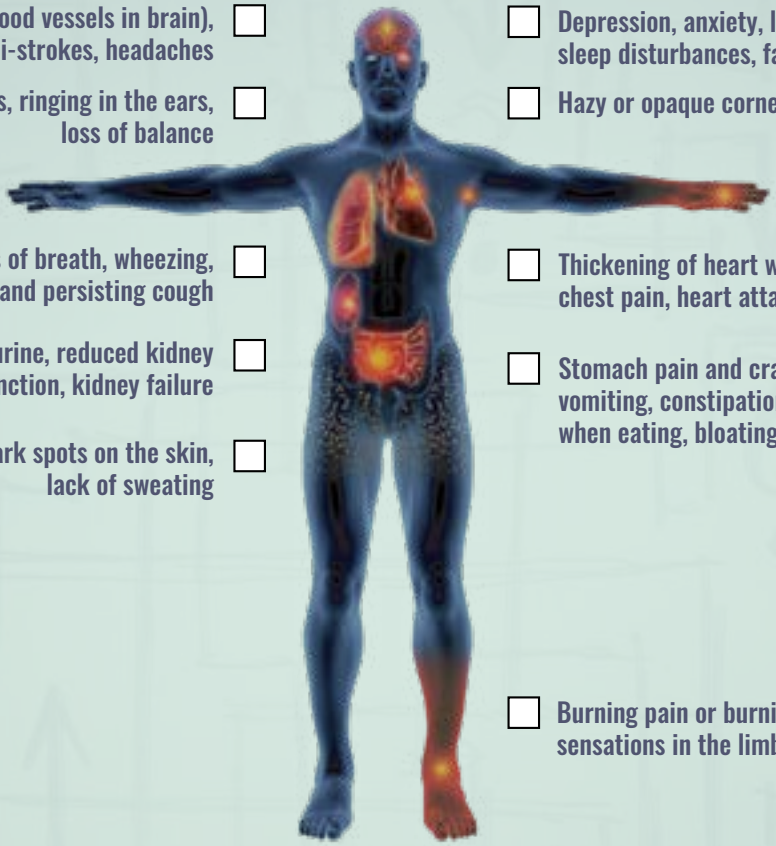
FABRY DISEASE SYMPTOM CHECKLIST

To be used in discussion with your doctor

Possible signs and symptoms of Fabry disease



- ☐ Strokes (clogged blood vessels in brain), mini-strokes, headaches
- ☐ Hearing loss, ringing in the ears, loss of balance
- ☐ Shortness of breath, wheezing, and persisting cough
- ☐ Protein in urine, reduced kidney function, kidney failure
- ☐ Small, dark spots on the skin, lack of sweating
- ☐ Depression, anxiety, low mood, sleep disturbances, fatigue
- ☐ Hazy or opaque cornea
- ☐ Thickening of heart wall, irregular heartbeats, chest pain, heart attack
- ☐ Stomach pain and cramping, diarrhea, vomiting, constipation, feeling full early when eating, bloating
- ☐ Burning pain or burning and tingling sensations in the limbs, heat intolerance

If you are experiencing any of these symptoms, print out this document and take it to your doctor for further information.

www.FabryDisease.ca

This section is for your doctor only

Fabry disease is a rare X-linked disorder that affects many parts of the body. It is commonly misdiagnosed.

Think of it when you see:

- Unexplained left ventricular hypertrophy (LVH)[3]
- Unexplained chronic kidney disease[2,4]
- Irregular heartbeats[2]
- Stroke or transient ischemic attack (TIA) in patients under 55[2,4]
- Pain or tingling in the limbs[4,5]
- Stomach pain, cramping, diarrhea, constipation, bloating[2,4]

Fabry disease is caused by a **mutation** in a gene called GLA. This gene provides instructions for making an enzyme called alpha-galactosidase A, or alpha-GAL A, which breaks down a fatty substance. A person who inherits this mutation has no or partially reduced activity of GAL A, resulting in a build-up of fatty substances in cells. The build-up damages the cells and organs, eventually leading to the signs and symptoms of Fabry disease.[1,3]

Please refer your patients to a nephrologist, geneticist or cardiologist, if you think they could benefit from a consultation for their diagnosis or treatment.

References: 1. National Institutes of Health, Genetics Home Reference. Your Guide to Understanding Genetic Conditions: Fabry disease. Available at: https://ghr.nlm.nih.gov/condition/fabry-disease# **2.** Mehta *et al. European Journal of Clinical Investigation.* 2004; 34:236-42 **3.** Germain. *Orphanet J Rare Dis* 2010;5:30 **4.** Mehta *et al.* Fabry Disease : Perspectives from 5 years of FOS. 2006. Avalable at : https://www.ncbi.nlm.nih.gov/books/NBK11586/ **5.** Colomba *et al. Oncotarget.* 2018; 9(8): 7758-62

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