# Fabry disease and the heart

**CPD Questionnaire**

**EARN FREE CPD POINTS**

Complete, scan and email to **cpd@denovomedica.com**. Alternatively, you can complete this questionnaire online at **www.denovomedica.com**. Fill in your details using **clear block letters** and mark the answers with a tick (✓).

**I agree that my CPD-accredited certificate will be forwarded to my e-mail address.** _____
(Signature of healthcare professional)

**(Online)**

| | | | |
|---|---|---|---|
| **First Name** | | **Profession** | |
| **Surname** | | **Telephone** | |
| **HPC No.** | | **City** | |
| **E-mail** | | | |

**1. Which statement is correct? FD:**

☐ **A** Is a lysosomal storage disorder
☐ **B** Arises from a deficiency of the α-Gal A enzyme
☐ **C** A and B

**2. Accumulation of Gb3 and related glycolipids in the lysosome is thought to have which of the following effects?**

☐ **A** Cytotoxic ☐ **B** Proinflammatory ☐ **C** Profibrotic ☐ **D** All of the above ☐ **E** B and C only

**3. Which statement is incorrect? Glycosphingolipid accumulation occurs in cells of the:**

☐ **A** Vascular system ☐ **B** Heart ☐ **C** Kidney ☐ **D** Nervous system ☐ **E** Muscle

**4. FD is caused by mutations in the ____ gene:**

☐ **A** GLA ☐ **B** β-Gal A ☐ **C** A and B

**5. Females with FD are more uniformly affected whereas males range from being asymptomatic to being severely affected:**

☐ **A** True ☐ **B** False

**6. Which statement is incorrect?**

☐ **A** There are over 965 reported mutations responsible for FD
☐ **B** Patients with the type 1 phenotype generally have < 15% of normal enzyme activity
☐ **C** Type 2 FD is a less severe form of the disease

**7. In patients with type 1 FD, unexplained LVH is due to:**

☐ **A** Myocyte hypertrophy ☐ **B** Glycolipid deposition ☐ **C** A and B

**8. Which statement is incorrect? In patients with type 1 FD:**

☐ **A** Patients frequently have minor thickening of the heart valves
☐ **B** Severely narrowed intramural coronary arteries are often surrounded by myocardial fibrosis
☐ **C** Females are at greater risk of ischaemic or haemorrhagic strokes

**9. Accurate diagnosis of FD in heterozygous females requires demonstrating:**

☐ **A** α-Gal A enzyme deficiency
☐ **B** The specific *GLA* gene mutation
☐ **C** A and B

**10. The ERTs, agalsidase alpha and agalsidase beta, have been demonstrated to prevent or slow the decline of end-organ damage associated with FD, especially if initiated early:**

☐ **A** True ☐ **B** False

**11. Which of the index patient's symptoms could alert the clinician to possible FD?**

☐ **A** The frequent ectopic beats
☐ **B** Multi-vessel disease on angiography
☐ **C** Severe LVH

**12. What other tests would you initiate in this index patient?**

☐ **A** CMR scan
☐ **B** Repeat echocardiogram
☐ **C** Genetic screening
☐ **D** A and C

**13. Which of this index patient's immediate family would you regard as a priority to screen?**

☐ **A** His son ☐ **B** His daughter

**14. What improvements would you expect in the index patient when using ERT?**

☐ **A** Improvement in LV function
☐ **B** Reduction in myocardial hypertrophy
☐ **C** Both of the above

**15. Should treatment of the index patient continue for life?**

☐ **A** Yes ☐ **B** No



© 2021 deNovo Medica