Fabry disease and the kidney

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1. FD is:

- ☐ **A** A disease caused by antibodies to fat
- ☐ **B** An abnormality of glucose metabolism
- ☐ **C** A lysosomal storage disease
- ☐ **D** A figment of our imagination

2. Which of these statements about FD are true?

- ☐ **A** It is an autosomal recessive disorder
- ☐ **B** It is an X-linked disorder
- ☐ **C** It is an autosomal dominant disorder
- ☐ **D** It is not inherited

3. A man who has FD:

- ☐ **A** Will transmit it to all his sons
- ☐ **B** Will transmit it to all his children
- ☐ **C** Will not transmit it to his children
- ☐ **D** Will transmit it to all his daughters

4. Which of the following is not true about FD?

- ☐ **A** It may be underdiagnosed
- ☐ **B** Females cannot be affected
- ☐ **C** Kidney disease is common
- ☐ **D** It can cause strokes

5. Which statement is incorrect? Classic FD:

- ☐ **A** Occurs only in males
- ☐ **B** Is often diagnosed earlier
- ☐ **C** Is associated with >1 % enzyme activity
- ☐ **D** Does not reduce lifespan

6. Which of the following is not a manifestation of FD?

- ☐ **A** Angiokeratoma
- ☐ **B** Corneal opacity
- ☐ **C** Hypohidrosis
- ☐ **D** Ash-leaf spot

7. Which statement is correct? Renal involvement in FD:

- ☐ **A** Presents in the second decade of life
- ☐ **B** Requires removal of both kidneys
- ☐ **C** Can be reversed if already established
- ☐ **D** Can be prevented

8. Which is not a feature on kidney biopsy of FD?

- ☐ **A** Vacuolated parietal and visceral epithelial cells
- ☐ **B** Zebra bodies on electron microscopy
- ☐ **C** Focal segmental glomerular lesions
- ☐ **D** Immune complex deposits

9. Which *GLA* gene abnormalities are not associated with late-onset disease?

- ☐ **A** E66Q
- ☐ **B** D313Y
- ☐ **C** M296I
- ☐ **C** R49P

10. FD should be screened for in which of the following instances?

- ☐ **A** Unexplained stroke in a young patient
- ☐ **B** Family history of death at a young age from cardiac disease
- ☐ **C** Heat, cold and exercise intolerance
- ☐ **D** All of the above

11. Screening for FD in females is done by:

- ☐ **A** Testing enzyme levels
- ☐ **B** Testing Gb3 in serum
- ☐ **C** Gene sequencing
- ☐ **D** Testing serum lyso-Gb3

12. Screening for FD in males is done by:

- ☐ **A** Testing Gb3 levels in serum
- ☐ **B** Cardiac endomyocardial biopsy
- ☐ **C** Renal biopsy
- ☐ **D** None of the above

13. Which statement is true with regard to treatment options for patients with ESKF and FD?

- ☐ **A** Peritoneal dialysis is not indicated
- ☐ **B** Transplantation is contraindicated without simultaneous liver transplant
- ☐ **C** Dialysis and transplantation can both be undertaken where possible
- ☐ **D** Late treatment can reverse the disease

14. Which statement is true of late-onset FD?

- ☐ **A** Treatment offers no benefit to the patient if diagnosed
- ☐ **B** The patient may present with kidney failure in the absence of classic features of FD
- ☐ **C** It is easy to diagnose

15. Treatment for FD:

- ☐ **A** Should be initiated early to prevent long-term complications
- ☐ **B** Involves ERT
- ☐ **C** Is costly
- ☐ **D** All of the above

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