Quiz

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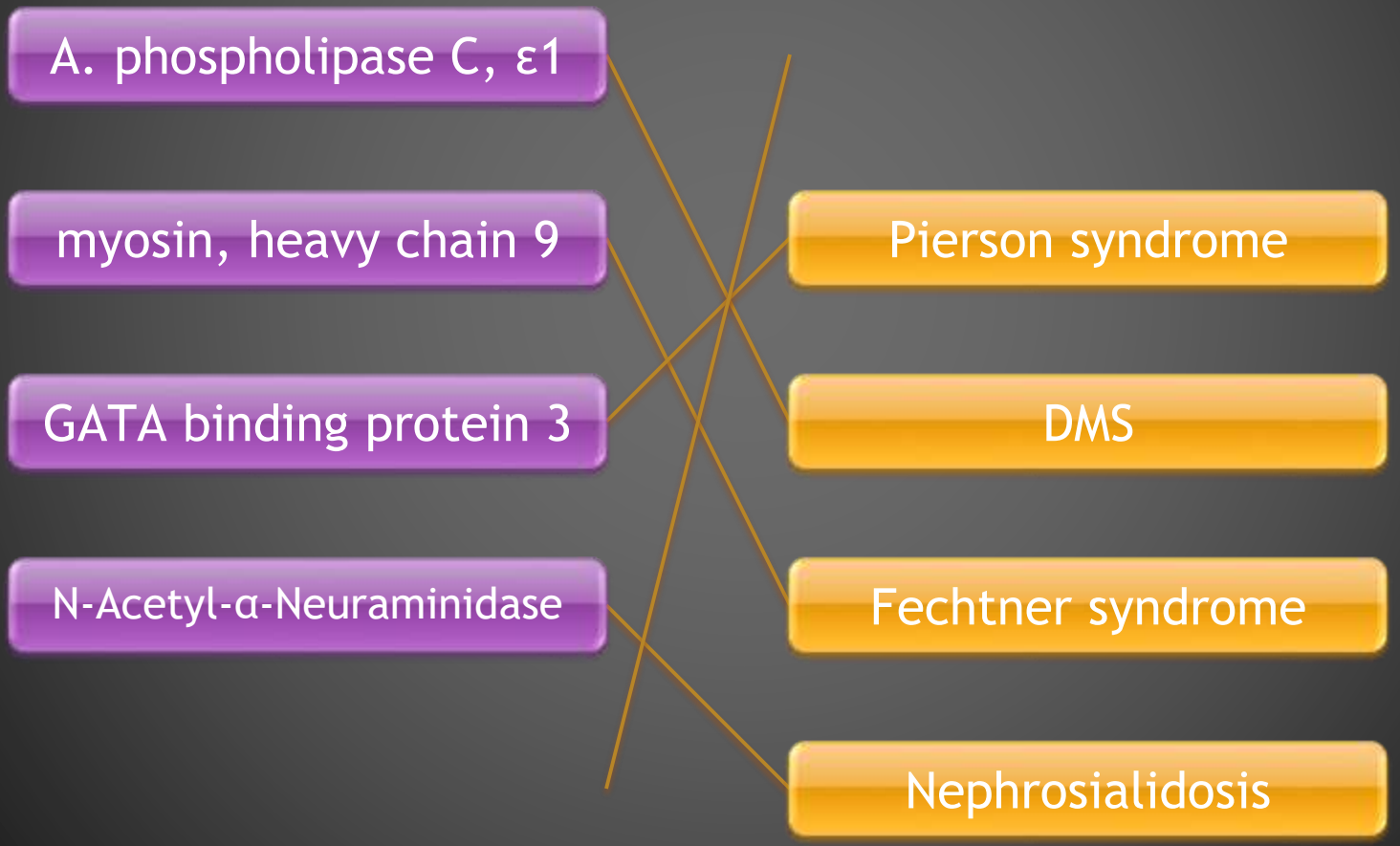
2. Which of the following is wrongly matched?



GATA binding protein 3 mutation

- Hypoparathyroidism, sensorineural deafness and renal disease, also known as HDR syndrome
- First described by Barakat et al. in 1977.
- Patients usually present with hypocalcaemia, tetany, or afebrile convulsions at any age.
- Hearing loss is usually bilateral and may range from mild to profound impairment.
- Renal disease includes nephrotic syndrome, cystic kidney, renal dysplasia, hypoplasia or aplasia, pelvicocalyceal deformity, vesicoureteral reflux, chronic renal failure, hematuria, proteinuria and renal scarring.
- Inheritance mode is probably autosomal dominant.
- This syndrome is primarily caused by haplo-insufficiency of the dual zinc finger transcription factor, GATA3

Pierson syndrome

- An autosomal recessive disease due to mutation in *LAMB2* which encodes the laminin β2 chain.
- Accounts for about 2.5% of nephrotic syndrome within the first year of life, usually manifested clinically within the first 3 months.
- Renal manifestations are accompanied by neurodevelopmental abnormalities
- Patients with the most severe manifestations die within the first year of life, while those with less severe manifestations progress to chronic kidney failure by 10 years of age and then require transplantation

Nephrosialidosis

- Sialidosis is an autosomal recessive disorder resulting from deficiency of α-neuraminidase, an enzyme that removes terminal sialic acid residues
- Infantile sialidosis is often recognized in the first year of life, and severely affects the kidney, causing symptomatic renal disease, which has been termed nephrosialidosis
- The basic morphological change of the kidney is the existence of membrane-bound vacuoles in almost all renal cells

WINNER

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CONGRATULATIONS

Thanks