CLINICAL QUIZ



Low molecular weight proteinuria, congenital myopia and hearing loss in a 10-year-old boy: Questions

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Case presentation

A 10-year-old boy was admitted to our hospital with the complaints of persistent proteinuria for 9 years. Foamy urine was found at the age of 1 year old. Some examinations had been performed at a hospital 9 years ago: urine tests were positive for urinary protein (+ ~ + +); 24-h urine protein quantity was 731.8 mg; urinary protein analysis revealed low molecular weight proteinuria (LMWP). Chinese medicine was given to the boy orally but had poor effects, and no other drugs were applied. Repeated routine urine tests showed persistent proteinuria.

The boy was diagnosed with congenital myopia at birth. Retinal detachment occurred in both eyes at the age of 3 years. Ophthalmologic surgery was performed, but his visual acuity was only 0.1. Hearing loss was found 1 year prior when he was 9 years old, and his hearing deteriorated over time during the previous 6 months. His psychomotor and speech development were normal. There was no family history.

Physical examination revealed widely spaced eyes, lowset posteriorly angulated ears, and enlarged globes leading to the appearance of prominent eyes.

The answers to these questions can be found at https://doi.org/10. 1007/s00467-023-05883-4.

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Urine tests were positive for urinary protein (++); 24-h urine protein quantity was 2100 mg (48.8 mg/kg). Repeated urinary protein analysis showed LMWP and mild to moderate elevated urinary immunoglobulin, transferrin and albumin levels. Ophthalmic examination revealed corneal opacity in the right eye and primary cataracts in the left eye. Based on the otolaryngology examination, moderate sensorineural hearing loss had occurred in both ears, and the boy was suggested to wear hearing aids.

Questions

- 1. What is the differential diagnosis?
- 2. What would you expect to see from kidney biopsy of the boy?
- 3. Which additional examination would be adopted for the diagnosis?
- 4. What is your diagnosis?

Data availability The data used and/or analyzed during the current study are available from the corresponding author on reasonable request.

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