

Dent's disease type 1 in a boy with severe hyperopia and mental dysfunction: a case report

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ABSTRACT

Dent's disease is a rare X-linked recessive proximal tubulopathy. It is typically characterized by low-molecular-weight (LMW) proteinuria, hypercalciuria, nephrocalcinosis, nephrolithiasis, hypophosphatemia, rickets and slowly progressive renal failure. The laboratory and clinical features may occur in various combinations. The early diagnosis of Dent's disease is often problematic because affected children may have mild clinical and biochemical signs, detecting LMW proteinuria

is not available in many laboratories, and genetic results are not clear in all cases. We report on a 12-year-old boy with Dent's disease type 1, severe hyperopia, and psychological dysfunction. To the best of our knowledge, he is the first patient with mutation in *CLCN5* gene and extrarenal symptoms described so far.

Key words: Dent's disease, low-molecular-weight proteinuria, *CLCN5* gene, hypercalciuria, nephrocalcinosis, hyperopia
