A girl presenting with gradually elevated levels of serum creatinine: Questions

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Case report

A 3-year-old girl was referred to our hospital due to suspected renal dysfunction based on the concentration of serum creatinine. She had regularly attended a regional hospital for follow-up for Down syndrome. During the follow-up period, She was never diagnosed with an underlying disease, such as congenital heart disease, hematologic disease, neuromuscular disorder, or renal disease. She took no medication and did not have a family history of renal disease. Her developmental milestones, especially motor developmental milestones, were delayed due to Down syndrome; she achieved head control at 8 months, sat up without support at 12 months, and took her first step at 32 months. Her DQ score from The Enjoji Scale of Infant Analytical Development Test was 48 at that time.

At the age of 3 years, elevated levels of serum creatinine were incidentally found. After this finding, her serum creatinine level gradually increased over 6 months, so she was referred to our hospital for assessment of renal dysfunction.

Her height was 80.2 cm (−4.9 SD for normal Japanese girls), and her body weight was 10.5 kg (−2.4 SD). Her blood pressure was within the normal range (77/43 mmHg). Physiological examination showed no abnormalities, such as heart murmur, edema, exanthema, hepatomegaly, splenomegaly, or hearing loss.

Biochemical analysis showed elevated levels of serum creatinine (1.03 mg/dL; estimated glomerular filtration rate (eGFR) by serum creatinine, 27 mL/min/1.73 m²) and blood urea nitrogen (24 mg/dL), with normal levels of serum cystatin C (0.82 mg/L; eGFR by cystatin C, 119 mL/min/1.73 m²), serum β₂-microglobulin (1.65 mg/L; eGFR by serum β₂-microglobulin, 99 mL/min/1.73 m²), electrolytes (sodium, 137 mEq/L; potassium, 4.4 mEq/L; chloride, 104 mEq/L; calcium, 9.4 mg/dL; magnesium, 2.0 mg/dL), serum total protein (6.6 g/dL), and serum albumin (4.1 g/dL). Serum immunoglobulins and complement were also normal (IgG, 931 mg/dL; IgA, 101 mg/dL; IgM, 124 mg/dL; IgE, 124 mg/dL; C3, 88 mg/dL; C4, 19 mg/dL; CH50, 38.0 U/mL). Myeloperoxidase (MPO)-antineutrophil cytoplasmic antibody (ANCA), Proteinase 3 (PR3)-ANCA, and anti-glomerular basement membrane (GBM) antibody were all negative. Complete blood counts revealed a normal level of hemoglobin (14.3 g/dL). There were no urinary abnormalities (urine red blood cells, <1/HPF; urine white blood cells, <1/HPF; urine total protein, <10 mg/dL; urine creatinine, 133 mg/dL).

Renal ultrasonography revealed no abnormalities, except for low-grade hydronephrosis in the right kidney.

Question

1. What would be your differential diagnosis of her elevated levels of serum creatinine?
2. What additional medical interview and examination would you do?

The answer to this question can be found at <https://doi.org/10.1007/s00467-020-04479-6>.

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Compliance with ethical standards

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