

선천성 호중구 감소증과 동반된 아밀로이드 침착증 1례

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A Case of Systemic Amyloidosis Associated with Cyclic Neutropenia

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Cyclic neutropenia is a rare disease characterized by regular cyclic fluctuation in the numbers of neutrophils. Patients with this disease suffer from recurrent infections at a regular interval of three weeks. Although cyclic neutropenia is benign in general, few patients develop reactive amyloidosis due to recurrent infections.

In summer 2007, a 14-year-old girl visited a hospital because of a huge palpable goiter and she was diagnosed with chronic thyroiditis and followed up without medication. In Aug. 2008, she was admitted to the hospital due to colitis. At that time, her thyroid function test revealed hyperthyroidism and she began to take propylthiouracil. On Oct. 2008, she was transferred to another hospital due to neutropenia. PTU was discontinued and synthroid medication was started to reduce huge goiter. On Dec. 2008, she developed mesenteric lymphadenitis with recurrent neutropenia. At that time, proteinuria was detected incidentally. On Feb. 2009, she was transferred to our hospital due to azotemia.

On admission, she has diffuse fixed hard huge goiters on physical examination. Her initial lab findings showed neutropenia (WBC 2,480, ANC 62), azotemia (BUN/Cr 10.4/1.2 mg/dL), proteinuria (1+ by dipstick), euthyroid function (T3 132 ng/dL, fT4 1.46 ng/dL, TSH 3.67 uIU/mL), negative autoimmune titer (C3/C4 110/25 mg/dL, Anti ds-DNA/RF/ANA/ANCA (-/-/-/-)). BM, thyroid and renal biopsies were done to rule out malignancy or autoimmune disease. However, the results of biopsies showed maturation arrest of myeloid cells, renal and thyroid amyloidosis. Echocardiography also suspected myocardial wall thickening with whitish deposition of amyloid fibrils. We checked serum protein- and immuno-electrophoresis, serum amyloid A, tuberculosis infection evidence for distinction between amyloid AA or AL. Serum PEP and IEP didn

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