

## 제1형 신경섬유종 환자에서 발생한 루푸스 신염

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### Lupus Nephritis in a Patient with Neurofibromatosis-1

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**Introduction :** Neurofibromatosis type 1 (NF-1) is a relatively common autosomal-dominant neurocutaneous disorder that affects ectodermal and mesodermal tissues. Systemic lupus erythematosus (SLE) is an autoimmune disease with diverse clinical manifestation and associated with pathogenic autoantibodies and genetic factor. SLE in a patient with neurofibromatosis has rarely been reported. We present a 25-year-old woman with generalized edema and arthralgia, who in addition had typical lesions of neurofibromatosis.

**Case :** A previously healthy 25-year-old woman who was diagnosed with NF-1 at 6 years ago presented with generalized edema. She complained arthralgia at both knee and hand joints and photosensitivity. The physical examination revealed numerous café-au-lait patches and multiple cutaneous neurofibroma on the trunk and lower extremities. Laboratory test showed leukocytes 4,820/mm<sup>3</sup> (segmented neutrophils 70.5%, lymphocyte 19.7%), hemoglobin 8.1 g/dL, platelet 80,000/mm<sup>3</sup> and erythrocyte sedimentation rate 115 mm/hr. Blood chemistry results were protein 6.9 g/dL, albumin 3.1 g/dL, C-reactive protein 1.9 mg/dL and BUN/creatinine 13.2/0.6 mg/dL. There were no significant findings for electrolytes. Proteinuria was (++++) at urinalysis. The sediment contained 30 to 49 red cells and 20 to 29 white cells per high power field. Antinuclear antibodies were 1:1280 and complement level of C3 was 28 mg/dL (normal: 90-180) and C4 <6.0 mg/dL (normal: 10-40). Ro and Sm antibodies were positive and La antibody was negative. Anti-ds DNA antibody was above five thousand IU/mL. Based on ACR criteria, she was diagnosed with SLE and lupus nephritis.

Two days after admission, we extracted kidney tissue from her left kidney for biopsy. On the third hospital day, treatment with prednisone (60 mg daily) was performed. After three weeks, the result was diffuse lupus nephritis (class IV-G, A/C). The patient started receiving intravenous cyclophosphamide and continued prednisone medication.

**Conclusion :** It is not clear whether there is a strong relationship between SLE and NF-1 or just coincidence. However, these diseases have a pathogenetically common background; genetic factors, environmental factors, malignant change. The further studies of possible association between SLE and NF-1 seem to be needed.

**Key Words :** 신경섬유종, 루푸스 신염

Neurofibromatosis, Lupus nephritis