中文題目：案例報告: 藍色橡皮痣症合併紅斑性狼瘡

英文題目：Blue Rubber Bleb Nevus Syndrome (BRBNS) with concurrent systemic lupus erythematosus

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Abstract

Blue rubber bleb nevus syndrome (BRBNS, Bean syndrome) is a rare disorder characterized by numerous cutaneous and internal venous malformations of unknown cause. Gastrointestinal lesions are pathognomonic.

A 61-year-old woman was admitted because of recurrent gastrointestinal bleeding. The Esophagogastroduodenoscopy revealed hemangioma- like submucosal lesion on gastric body. Histopathological examination of the submucosal lesion showed venous malformation. Multiple blue elastic skin lesions on tongue, mouth, trunk, bilateral hands, and feet were noted. The histopathology of biopsy axillary mass was also compatible with venous malformation. Reviewing her previous colonoscopy, one resected polyp also showed venous malformation. Therefore, the diagnosis of BRBNS was confirmed.

She also presented with rapid progressed renal failure with proteinuria and received hemodialysis. A renal biopsy was performed and the histopathology showed focal segmental glomerulosclerosis, which was compatible with lupus nephritis. In addition, cognitive dysfunction, recurrent seizure, and progressive 4 limbs weakness developed after admission., Cerebrospinal fluid (CSF) analysis revealed elevated CSF IgG index (IgG index: 0.75). Brain MRI showed bilateral frontal and parietal lobes diffuse white matter edema. Nerve conduction study (NCS) revealed polyneuropathy. Systemic autoimmune disease with neurologic involvement was suspected. The serologic tests showed positive antinuclear antibodies, direct Coombs' test, and hypocomplementemia. Late-onset lupus with chronic nephritis and neurologic involvement was considered.

To date, only few cases of BRBNS were reported in Taiwan. Most of the cases were found in children, while. This is the second case of adult- onset BRBNS. Moreover, this patient has concurrent systemic lupus erythematosus (SLE) with nephritis and neurologic involvement. Treatments for BRBNS -related gastrointestinal bleeding include endoscopic resection and sirolimus. In this case, we present a good result of endoscopic resection. Moreover, hydroxychloroquine 100mg/day, azathioprine 25mg/day, and prednisolone 5mg/day were prescribed for treatment of SLE. Her cognitive dysfunction improved gradually and the frequency of hemodialysis

decreased from 3 times a week to 1 time a week.

In summary, we presented a rare case of adult-onset BRBNS with concurrent systemic lupus erythematosus. The treatment outcomes of both diseases are excellent, while the relationship between BRBNS and SLE need to be elucidated.