以脊髓病變為主要表現的自體免疫多腺體症候群：一病例報告

陳冠妃[1]　蔡宗璋[1]　許怡婷[1]　蔡崇豪[1,2]
中國醫藥大學附設醫院 [1]神經部 [2]神經醫學研究室

Polyglandular autoimmune syndrome presenting with myelopathy: a case report

Kuan-Fei Chen[1], Tzung-Chang Tsai[1], Yi-Ting Hsu[1], Chong-Haw Tsai[1,2]
Departments of [1]Neurology and [2]Neuroscience Laboratory, China Medical University Hospital, Taichung, Taiwan

Background　Vitamin B12 deficiency may present with pernicious anemia, macrocytic anemia, subacute combined degeneration of the spinal cord, peripheral neuropathy, and psychiatric symptoms. Here we report a case of polyglandular autoimmune syndrome related vitamin B12 deficiency

Case Report　A 40-year-old woman with a past history of hyperthyroidism complained of progressive general weakness and unsteady gait in recent one year. She has been a vegetarian since the age of thirteen. One year ago, she stated to notice loss of taste and smell sensation. Tingling and tight sensation of both lower extremities developed and then extended upward to the upper trunk within months. Progressive lower limb weakness was noted as well. She felt like standing on the cloud and walking like a robot. On presentation, she was found to have impaired attention and short-term memory. Neurological examination disclosed normal findings of cranial nerves. The motor strength of the upper limbs was 4/5 and the lower limbs 4-/5. Muscle tone increased as spasticity in both lower extremities. Deep tendon reflexes were normal in the upper and increased in the lower extremities. There were bilateral extensor plantar responses. Impaired vibration sensation and joint position below T4 level were determined. Cerebellar examination was normal but Romberg's sign was positive. Lhermitte's sign was absent. Laboratory tests revealed Hb 7.2 g/dL; MCV: 75.5 fL; Vitamin B12 level: 101 pg/ml (180–914); folic acid level: 14.94 ng/ml (>3); iron 173μg /dl (66-178); Serum TSH: 1.566μIU/ml (0.34-5.60); free T4: 0.81 ng/dl (0.54-1.40); Anti-microsomal antibody: 1:1600. Anti-parietal cell antibody was positive. The routine CSF analysis revealed normal findings. Genetic studies disclosed α-thalassemia. The nerve conduction studies disclosed symmetrical sensori-motor polyneuropathy with predominant axonal degeneration. The findings of somatosensory evoked potential were also suggestive of diffuse peripheral neuropathies. Brain and C-spine MRI were unremarkable. Thyroid echo revealed autoimmune thyroid disease.

Conclusion　In this case, although she is a vegetarian, anti-parietal cell antibody related vitamin B12 malabsorption should be the main cause of vitamin B12 deficiency. Vitamin B12 deficiency mostly causes macrocytic anemia, but the patient presented with microcytic anemia. We presume this is because the patient has thalassemia and the MCV might be lower before. In addition, besides pernicious anemia, autoimmune thyroid disease was also diagnosed. She might be a case of polyglandular autoimmune syndrome type 3.

論文分類：K 神經免疫學 D 神經肌病學 L 其他
通訊作者：陳冠妃
所屬醫院:中國醫藥大學附設醫院　神經部