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A Case of Charcot-Marie-Tooth Disease with Recurrent Guillain-Barré Syndrome

Dong Hwan Kim[1], Seung Ah Lee[1], Young Rok Han[1], Eo Jin Park[1*], Seung Don Yoo[1†]

Kyung Hee University Hospital at Gangdong, Department of Physical Medicine & Rehabilitation[1]

Guillain-Barré (GB) syndrome is clinically characterized by symmetrical weakness of the limbs as well as hyporeflexia or areflexia; it progresses to the serious condition within 4 weeks and causes acute flaccid paralysis. Charcot-Marie-Tooth (CMT) disease is a genetic disorder; patients with CMT disease present with a progression of chronic motor and sensory neuropathy. The lifetime prevalence of CMT disease is reported as 40/100,000 in worldwide. The case of CMT disease with recurrent GB syndrome is rare A 29-month-old girl was referred to our clinic for further evaluation and treatment of muscle weakness. On history taking, the patient had no past history of trauma, but being diagnosed with CMT disease in 2009 and GB syndrome in 2011 at other hospitals. Despite a lack of duplication of the peripheral myelin protein 22 (PMP22) gene, the patient showed findings that are indicative of peripheral motor and sensory polyneuropathy on electromyography (EMG). Furthermore, the patient also showed abnormal thyroid functions. After 2 injections of immunoglobulins in January and March, the patient achieved improvements in symptoms. In 2013, the patient developed general paresis accompanied by changes in respiration and voice formation as well as fever, for which a 5-day-course of immunoglobulins was administered. In the first week of July of 2013, the patient developed paralysis of both arms, thus being treated with immunoglobulins. Then, the patient received a decreased dose of prednisolone from 25 mg to 20 mg. In December of 2013, the patient developed general paresis involving the neck. The currently, the patient was taking prednisolone 20 mg in August of 2017, the patient was treated with immunoglobulins again, but showed no notable findings in the muscle weakness. At our institution, the patient presented with a delayed milestone, dysphonia, dysarthria, and EMG findings that are suggestive of peripheral motor and sensory polyneuropathy. On urological examination, the patient had urinary frequency, which is indicative of GB syndrome. Currently, the dose of prednisolone was maintained at 20 mg. According to a review of the literature, there is a relationship between primary hypothyroidism and GB syndrome; gangliosides are abundantly present in thyrocytes and neuronal cells. This may lead to the formation of auto-antibodies involved in the pathogenesis of GB syndrome. Moreover, it has also been reported that patients with CMT disease would be vulnerable to worsening of clinical and neurophysiological findings if they have comorbidities, such as diabetes, hypothyroidism, exposure to toxins and obesity. In conclusion, our case indicates that physicians should consider the possibility of thyroid dysfunction in patients with signs and symptoms that are suggestive of CMT disease and GB syndrome.