Case Report

Porphyria With Predominantly Involvement of Bilateral Facial Nerves

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Abstract

Porphyria is an uncommon disease caused by the deficient activity of a heme biosynthetic enzyme, which presents clinically with neurological and psychiatric symptoms. To our knowledge, this is the first report describing porphyria in a case with the predominant bilateral facial paralysis without clearly disclosing other already known neurological manifestations.

Keywords: Porphyria, facial diplegia

INTRODUCTION

Porphyria is an uncommon, autosomal dominant condition resulting from a relative deficiency of the enzyme porphobilinogen deaminase of the heme biosynthetic pathway. The clinical expression of the disease is characterized by acute, life-threatening attacks of porphyric neuropathy that include abdominal pain, motor and sensory neurologic deficits and psychiatric symptoms. The neurologic manifestations are usually those of a polyneuropathy, involving the motor nerves more severely than the sensory ones. An acute or subacute, motor-predominant, axonal polyneuropathy is characteristic of porphyria. Neuropathy occurs in about 50% of attacks (1). Prominent weakness is often seen symmetrically in extremity muscles, cranial nerve innervated muscles and rarely extraocular muscles. The symptoms may ascend and spread in a few days to the trunk. Symptoms include severe abdominal pain, vomiting, constipation, hypertension, tachycardia, and bladder dysfunction which have been ascribed to autonomic neuropathy (2,5).

CASE PRESENTATION

A 20 years old man developed a sudden, simultaneously bilateral facial weakness. He had had a flu-like infection, fatigue and back pain ten days prior to the onset of facial weakness on the two sides. There was no relevant past medical history. On examination he had complete bilateral facial paralysis of peripheral type. All the tendon reflexes were decreased in the upper limbs and unelicitable in lower extremities. There were no other neurological signs. Nerve conduction studies performed seven days after the onset of facial weakness revealed mildly slowed motor conduction velocities of right posterior tibial,
left peroneal, posterior tibial and ulnar nerves without conduction block as well as prolonged distal motor latencies. Soleus H reflexes were bilaterally unelicitable. Sensory nerve conduction velocities were normal. Facial nerves were bilaterally inexcitable. Needle electromyography of limb muscles was normal. Routine haematology, serum biochemistry including immunoglobulines and protein electrophoresis were normal. CSF glucose, protein and IgG levels and IgG index were within normal limits. No cells were seen in CSF microscopy and oligoclonal bands were negative. Cranial magnetic resonance imaging did not demonstrated any abnormality. Electroencephalography was normal. The patient was initially diagnosed as Guillain Barré Syndrome (GBS) and treated with intravenous immunoglobulin in a dosage of 0.4 g/kg of body weight per day for 5 consecutive days. During his hospital stay, a behavioral alteration characterized with agitation and increased irritability was observed and subsided in a few days. Because of sudden appearance of behavioral and mood changes porphyria was considered. Serum and urinary porphobilinogen was positive, serum uroporphrine was elevated (162,6 microgr per day) and coproporphrine was normal. On day 20, motor conduction velocity values of the tibial, peroneal and ulnar nerves returned to normal, while electromyographic studies disclosed totally loss of motor unit potentials with denervation potentials in bilaterally orbicularis oculi, orbicularis oris and frontal muscles.

DISCUSSION
Porphyria is an autosomal dominant condition resulting in a deficiency of porphobilinogen-deaminase, the cytosolic enzyme in the heme biosynthetic pathway that catalyses the condensation of 4 molecules of porphobilinogen by a series of deaminations to form the linear tetrapyrrole, hydroxymethylbilane, which is then enzymatically cyclized to form uroporphyrinogen 3 by uroporphyrinogen 3 synthase. There is feedback inhibition of the enzyme aminolevulinic acid-synthase by the end product, heme. Porphyria is characterized by genetic heterogeneity and highly variable expression (4). The clinical manifestations of porphyria are distinctive, and include recurrent attacks of acute abdominal pain with gastrointestinal symptoms (so-called neurovisceral episodes), neuropsychiatric symptoms, and an acute or subacute motor-predominant polyneuropathy. The neurological manifestations of porphyria are usually those of a polyneuropathy, involving the motor nerves more severely than the sensory ones. Equal involvement of both sensory and motor nerve fibers are rarely seen, and sometimes autonomic nerves are affected. The symptoms may begin in the feet and legs and ascend, or they may begin in the hands and arms and spread in a few days to the trunk and legs. Sensory loss, often extending to the trunk, is present in half of the cases. Facial paralysis, dysphagia, and ocular palsies simulating GBS, are the features, which seen in the most severe cases. Porphyric polyneuropathy must be differentiated from the other causes of the acute or subacute motor neuropathy. GBS is distinguished by the early loss of tendon reflexes, as well as the lack of prominent muscle wasting in the more common demyelinating form (2,3). Electrophysiological tests and cerebrospinal fluid protein serve to distinguish the one from the other. Our patient suffered a severe bilateral facial paralysis with acute onset. The diagnosis of GBS was initially considered. However, clinical symptoms including facial paralysis and mood changes suggested central nervous system involvement in addition to the peripheral nerve disorder, leading us to consider other diagnoses. Owing to high levels of porphyrine metabolites the diagnosis of porphyria was then accepted. In this case, the diagnosis of GBS was excluded due to absent of albumino-cytologic dissociation in CSF, presence of mildly slowed motor conduction velocities of motor nerves without conduction block, and the manifestations of central nerve system. The case that we report here is of particular interest because of having acute facial diplegia and trivial signs of peripheral nerve involvement (mild slowing in motor conduction velocities without conduction block) and mood and behavioral alterations.
(agitation, irritation and restlessness). Bilateral facial paralysis is rarely seen in porphyria but the incidence is not known. Facial diplegia as the first manifestation of porphyria has not been previously reported. This appears to be the first reported case of porphyria with the predominant bilateral facial paralysis without clearly disclosing the other already known manifestations of the disease.

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