ACUTE INTERMITTENT PORPHYRIA:
AN ATYPICAL PRESENTATION WITH POLYNEUROPATHY

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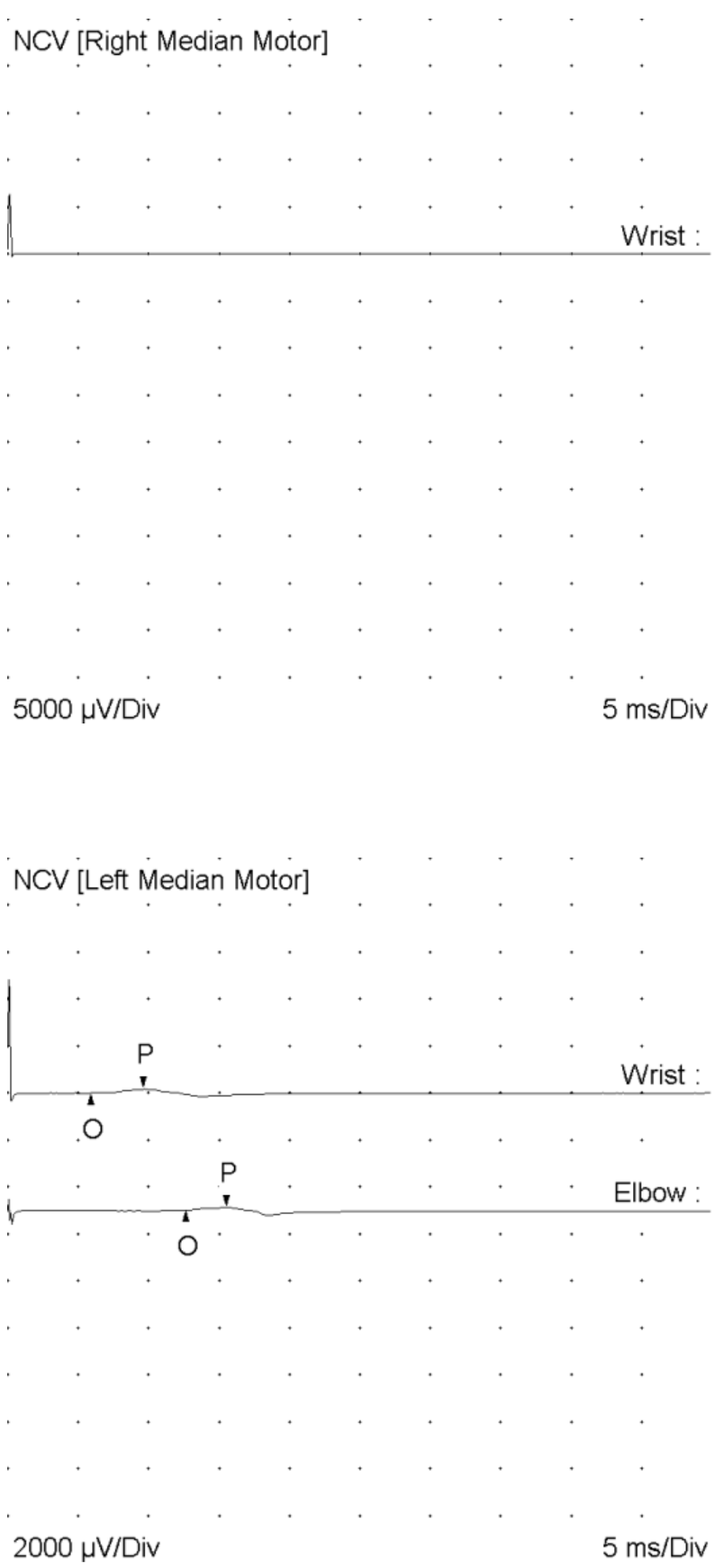

Figure 1. Absence of response right median motor nerve and very low amplitude in left median motor nerve.

INTRODUCTION

Porphyrias are rare genetic disorders caused by enzyme deficiency in heme biosynthesis. Its clinical presentation is varied and polyneuropathy is very rare so it can be a diagnostic and therapeutic challenge. We present the case of a patient with symptoms of acute abdominal pain accompanied by hematuria, who rapidly develop progressive weakness in all 4 limbs and severe dysphonia. Later diagnosis of acute intermittent porphyria (AIP) was confirmed.

PATIENT INFORMATION AND CLINICAL FINDINGS

A 30-year-old man with no significant history presented with abdominal pain initially treated as intestinal obstruction without improvement, then presented weakness in the upper limbs that progressed to the lower limbs. On physical examination generalized hyporeflexia, predominantly proximal weakness. Spine imaging showed bilateral and symmetrical thickening and enhancement of the thoracolumbar roots. Guillain-Barré Syndrome (GBS) was suspected without improvement with immunoglobulin. The electrodiagnostic study showed acute sensorimotor axonal polyneuropathy, due to changes in urine color, porphobilinogen compatible with porphyria was requested, intravenous hemantine was started without recovery in motor function.

DISCUSSION AND CONCLUSIONS

IAP has an incidence of 0.5-1 in 100,000 population, it rarely manifests with polyneuropathy, the presentation is like GBS, however, with a proximal and descending onset. In nerve conductions studies, findings are axonal compromise without evidence of conduction blocks or conduction slowing. Electromyography may be normal or show signs of denervation and reinnervation. Most patients have complete recovery, but a small group persists with chronic polyneuropathy despite treatment, with a poor prognosis for rehabilitation.

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