

신경근육재활 및 전기진단

게시일시 및 장소 : 10 월 18 일(금) 08:30-12:20 Room G(3F)

질의응답 일시 및 장소 : 10 월 18 일(금) 10:12-10:16 Room G(3F)

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A Case Report of Acute intermittent porphyria with a good prognosis

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Introduction

Porphyria is a complex and uncommon metabolic condition caused by deficiencies in the pathway of heme synthesis. Acute intermittent porphyria(AIP) is an autosomal dominant disorder due to deficiency of porphobilinogen(PBG) enzyme deficiency and it presents diverse group of symptoms making its early diagnosis difficult. We report a case of AIP which the diagnosis was delayed.

Case report

A 23-year-old female had an abdominal pain and visited emergency room (ER) at 11 Feb. 2018. Intussusception was suspected and laparoscopic exploratory laparotomy was done. Her symptoms relieved spontaneously so she discharged from hospital at 2018.02.24. She had a generalized tonic-clonic seizure in the day of discharge and revisited ER. She had four more GTC type seizure in ER. She was admitted to the Intensive Care unit in Department of Neurology. Magnetic resonance imaging(MRI) was performed which showed PRES(Posterior reversible encephalopathy syndrome). The GTC type seizure continued and she was treated with sedative agents and anti-epileptic drugs. Her motor weakness was not evaluated early, manual muscle test result in all limbs were grade P to F. The EMG result showed motor dominant polyneuropathy and clinicians suspected Guillain-Barre Syndrome at first. And because acute severe abdominal pain and neuropathy occurred same time clinicians suspected AIP so urine PBG test was done. PBG result was positive, and she was diagnosed as AIP. Although intravenous hemin therapy was applied her the motor weakness and respiratory paralysis did not recover. Intermittent positive pressure ventilation was applied after tracheostomy. She kept on rehabilitation for her motor weakness. While she was treated her abdominal pain started and she could not breath in and out fully even though ventilation was applied. 10% dextrose water relieved abdominal pain but was transient and dyspnea kept on. Urine PBG result were increased and AIP recurrence were suspected. She had four days of intravenous hemin treatment for AIP. Her abdominal pain disappeared and she kept on rehabilitation and wean out ventilator successfully in 2018.08.28. Her K-MBI(Korean modified barthel index) was 0 and FAC(Functional ambulation category) scale was 0 on 18.06.28. Follow up EMG showed regeneration evidence. The K-MBI score was 34 and FAC scale was 2 on 18.10.31. At the point of 19.07.01 her K-MBI score is 92 and FAC scale is 4.

Conclusion

Abdominal pain has a variety of possible causes. But abdominal pain with neurological symptoms should consider porphyria. It can be considered that toxicity of porphyria can cause various neurologic symptoms including peripheral neuropathy and PRES. In this case young female recovered from her severe neurological condition with an appropriate medical service and sufficient rehabilitation. Although further studies should be taken, it can be suggested that severe neurological involvement due to AIP can have good prognosis.