J Gastroenterol
https://doi.org/10.1007/s00535-020-01752-1

The Japanese Society of Gastroenterology

EDITORIAL

Diagnosis for Wilson disease: this disease may not be a rare disease

Masaru Harada[1]

Received: 9 November 2020 / Accepted: 11 November 2020

Keywords *ATP7B* · Ceruloplasmin · Genetic disease · Wilson disease

Wilson disease is an autosomal recessive genetic disorder of abnormal copper metabolism. Until now the estimated prevalence of Wilson disease has been considered about 1 in 30,000 [1, 2]. Previously, physicians sometimes disregarded this disease. Recently, many physicians recognize this disease. The diagnosis of Wilson disease is based on a combination of clinical features, serum ceruloplasmin concentration, urinary copper excretion, examination of Kayser-Fleischer ring, measurement of hepatic copper contents and genetic analysis of *ATP7B* [1–3]. There are some guidelines for the diagnosis of this disease and many patients are diagnosed without genetic analysis of *ATP7B* [1, 2]. Some patients may be excluded as different hepatic diseases without genetic analysis of *ATP7B*.

Examination of serum ceruloplasmin is important for the diagnosis of Wilson disease. However, serum ceruloplasmin may be low in some different conditions such as heterozygotes of *ATP7B* mutations, renal diseases, severe end-stage live diseases or aceruloplasminemia [4, 5]. In addition, serum ceruloplasmin may be normal in some patients with Wilson disease [6, 7]. Measurement of hepatic copper content has been considered the best biochemical examination for the diagnosis for Wilson disease [1, 8]. However, the distribution of copper in the liver is sometimes inhomogeneous [1]. It is sometimes elevated in other liver diseases such as chronic cholestatic conditions [6]. Especially, some patients with idiopathic copper toxicosis were misdiagnosed as Wilson disease due to the extensively elevated hepatic copper content [9, 10].

In this issue of the *Journal of Gastroenterology*, Garcia-Villarreal et al. investigated the method for the early diagnosis of Wilson disease [11]. They found a combination of measurement of serum ceruloplasmin with genetic analysis of *ATP7B* was useful for the definitive diagnosis for Wilson disease. Clinical manifestations vary in patients with Wilson disease. There are hepatic, neural and other organ manifestations. Clinical features usually appear during the second or third decades. Therefore, the early diagnosis is sometimes difficult in children and patients with mild manifestations. However, the mutation of *ATP7B* is present from birth in patients with Wilson disease. Considered with these problems for the diagnosis of Wilson disease, a combination of serum ceruloplasmin and genetic screening of *ATP7B* is very useful, although it takes some time to obtain the result. Therefore, it may be difficult to use emergency cases. However, it must be useful for non-typical cases and asymptomatic patients. Therefore, the prevalence of this disease must be greater than the previous estimate considered from a recently published report [12]. Using this method presented by Garcia-Villarreal et al. [11] in this issue, we may be able to diagnose patients who were not diagnosed previously as Wilson disease. In additions, we will be able to eliminate the patient with idiopathic copper toxicosis who is sometimes misdiagnosed as Wilson disease [10].

Wilson disease is a rare genetic metabolic disorder that we can treat using several drugs. Therefore, a precise diagnosis is necessary using adequate methods for patients

✉ Masaru Harada
msrharada@med.uoeh-u.ac.jp

[1] Third Department of Internal Medicine, School of Medicine, University of Occupational and Environmental Health, 1-1 Iseigaoka, Yahatanishi-ku, Kitakyushu 807-8555, Japan

Published online: 03 January 2021

with this disease and this disease may not be a rare genetic metabolic disease.

Compliance with ethical standards

Conflict of interest The author declares no conflict of interest.

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