Introduction

The true prevalence of Wilson’s disease (WD), remains unknown. The estimated genetic prevalence in the UK (142/million) is higher than the clinical prevalence (15/million) reported in other European studies. We aimed to (1) estimate the prevalence of WD in Nottingham, (2) assess the utility of readily available laboratory and clinical data to identify patients with WD, and (3) propose a system to identify patients with WD nationally.

Methods

Patients with WD attending Nottingham University Hospital (NUH) 2011–2018 were identified using multiple sources of case ascertainment: (1) serum ceruloplasmin level <0.2 g/l (2) 24-hour urinary copper measurement (3) ‘Wilson’ in a liver biopsy report (4) hospital prescription for Penicillamine, Trientine or Zinc and (5) admission to NUH coded with ICD-10 Code E83.0 ‘Disorder of copper metabolism’. We identified potential cases of WD using the Leipzig score, confirmed the diagnosis in hospital records and calculated the point prevalence with Poisson confidence intervals using the Office for National Statistics mid-2017 population estimates for the denominator population.

Results

We identified 1,794 patients from ≥1 source, and 13 patients had WD. The overall prevalence of WD was 12.6/million (95%CI 6.7–21.6); males 15.6/million (95%CI 6.7–30.8) and females 9.6/million (95%CI 3.1–22.5). Patients with confirmed WD were followed up by: Hepatology 12 (92.3%), Neurology 6 (46.2%), Psychiatry 4 (30.8%), and Renal 1 (7.7%). 5 (38.5%) were being managed for cirrhosis secondary to WD. 1 (7.7%) had received a liver transplant. Additionally, 23 (1.3%); males n=19 patients had a low (<0.2 g/l) serum ceruloplasmin level and an elevated 24-hour urinary copper, but had not been investigated further for WD. These patients, if confirmed to have WD, would increase the prevalence of WD to 34.9/million (95%CI 24.5–48.4).

Discussion

This is the first UK population-based study of WD prevalence. The prevalence found in this study is lower than the previous UK population-based genetic study, but comparable to European population-based clinical studies. The significant difference in prevalence between genetic and clinical studies may be due to under-diagnosis of WD or variable genetic penetrance. The lower prevalence of WD among females than males. The method of case ascertainment used in this study may be a cost-effective method of identifying patients with WD, and similar practices could be adopted nationally.