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ORIGINAL ARTICLE

Characteristics and prevalence of Wilson's disease: A 2013 observational population-based study in France

Aurélia Poujois[a], **France Woimant**[a], **Solène Samson**[b], **Pascal Chaine**[a], **Nadège Girardot-Tinant**[a], **Philippe Tuppin**[b,*]

[a] *Département de Neurologie, Centre National de Référence pour la Maladie de Wilson, Hôpital Lariboisière, Assistance publique–Hôpitaux de Paris, 75010 Paris, France*
[b] *Direction de la stratégie des études et des statistiques, Caisse Nationale d'Assurance Maladie, 50, avenue du Professeur-André-Lemierre, 75986 Paris cedex 20, France*

KEYWORDS
Wilson's disease;
Zinc;
Epidemiology;
Geographical distribution;
Healthcare use;
D-penicillamine

Summary
Background and aims: Only a few epidemiological studies on the incidence and prevalence of Wilson's disease (WD) have been performed to date, and the results vary widely according to the reports. The aim of the study was to investigate the prevalence, ambulatory care and treatments of patients with WD in France.
Methods: Among the 58 million general health scheme beneficiaries (86% of the French population), people managed for WD in 2013 were identified using hospitalisation diagnosis in 2011–2013 or specific long-term disease status with a 100% reimbursement for specific healthcare in 2013. Data were derived from the Sniiram (National Health Insurance Information System database). Prevalence by age and sex were calculated.
Results: In 2013, 906 prevalent cases were identified, yielding a crude prevalence of 1.5 cases per 100,000; 1.65 per 100,000 in males and 1.44 per 100,000 in females. This prevalence is comparable to that reported in other population-based studies in European countries and to a study using a similar method. Almost 40% of patients were treated by D-penicillamine and 14.3% were treated by zinc acetate. Trientine, delivered on a compassionate basis, was not available in the reimbursement database. In 2013, 1.3% of patients underwent liver transplantation and 4% had already undergone liver transplantation in previous years. Fifteen per cent of patients received antidepressants, a higher rate than in general population.
Conclusions: This is the first French population-based epidemiological study of WD in a comprehensive population based on administrative data and constitutes an important step to understand the impact of WD and to study quality of care.
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* Corresponding author.
E-mail address: philippe.tuppin@cnamts.fr (P. Tuppin).

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Introduction

Wilson's disease (WD) is a rare autosomal recessive disorder affecting copper metabolism, characterized by accumulation of copper in the liver and brain. WD is due to mutations in the *ATP7B* gene, predominantly expressed in the liver [1,2]. Defective ATP7B function impairs both copper incorporation in caeruloplasmin and copper release into the bile, resulting in accumulation that leads to cell death [3]. In the absence of diagnosis and treatment at this stage, hepatic free copper spills into the bloodstream and is deposited in other organs such as the brain [4]. WD consequently evolves towards a systemic disease and its clinical features can vary from an asymptomatic state to hepatic and neuropsychiatric manifestations.

Only a few nationwide epidemiological studies on the incidence and prevalence of WD have been performed to date, and incidence and prevalence rates have changed over recent years. Early studies suggested that the prevalence of WD was about 30 per million inhabitants [5], but subsequent epidemiological studies have suggested a lower prevalence in European countries with a low degree of consanguinity, estimated at between 12 and 20 per million inhabitants [6]. In a recent UK population-based genetic study, the frequency of individuals predicted to be carrying two mutant pathogenic ATP7B alleles was 1/7026, which is considerably higher than the estimated prevalence of WD [7]. Apart from disease registry or population-based studies restricted to limited geographical regions, only one nationwide population-based study on WD prevalence was performed in Taiwan, using a health insurance scheme database [8–12]. Nevertheless, determining the nationwide epidemiology, sociodemographic characteristics of WD patients and their health care utilisation constitutes an important step towards a better understanding of the public health impact of this disease and how WD patients are treated.

We conducted a nationwide population-based study using the French national health insurance information system database to estimate the prevalence of WD based on healthcare reimbursement data and provide information on health care utilisation by WD patients in 2013.

Patients and methods

Data source and population

The French National Health Insurance information system (Système national d'information interrégimes de l'Assurance maladie [Sniiram]) database comprehensively and individually records all outpatient prescriptions and healthcare procedures reimbursed to beneficiaries of the various French health schemes [13], but does not comprise any clinical information concerning the results related to consultations, prescriptions or examinations. However, attribution of long-term disease (LTD) status for a severe and costly chronic disease e.g. Wilson's disease, validated by a national health insurance physician at the request of the attending physician, allowing exemption of co-payment, can provide information about the nature of the diseases treated. An anonymous and unique identification number for each beneficiary allows this information to be linked to the data collected by the PMSI (Programme de médicalisation des systèmes d'information) [Medical Information System Programme] during hospital stays in the various types of health care institutions in France. Hospital diagnoses are coded according to the International Classification of Diseases 10th edition (ICD-10), in the same way as the diagnoses allowing attribution of LTD status. Therefore, all French residents seeking outpatient health care or inpatient care are captured by the Sniiram. Vital status and date of death are available, but not the cause of death. For data protection reasons, Sniiram data are currently available for a three-year period in addition to the current year.

The national health insurance general health scheme covered about 76% of the 66 million inhabitants of France in 2013, as well as various population groups covered by local mutualist sections for students, civil servants, etc. (i.e. 86% of the French population). The Mutualité sociale agricole (agricultural workers' health insurance fund) and the Régime social des indépendants (self-employed health insurance fund) each cover 5% of the population, and the remaining 4% is covered by other schemes. The population of the present study was therefore composed of general health scheme beneficiaries, including local mutualist sections (58 million inhabitants). Beneficiaries of the other funds were not included due to the lack of comprehensive data for LTD status in 2013.

Case definitions and data analysis

We applied three alternative definitions when identifying WD cases in 2013. First, we identified all LTD with ICD-10 code E83.0 for copper metabolism disorders. Second, we used the 2011, 2012 and 2013 hospital discharge diagnoses to identify all individuals discharged from hospital at least once with a principal, related or associated diagnosis ICD-10 code E83.0 for copper metabolism disorder or the ''Orphanet'' WD code (MR905) over this three-year period. Third, we applied a composite definition according to which patients were considered to have WD when they complied with either the first or second definition. Information regarding comorbidities was obtained using other associated LTD diagnoses. WD drug therapies were defined by at least one reimbursement in 2013 for D-penicillamine or zinc acetate, as well as antidepressant medications. Information on Trientine, which is available in France under a compassionate use programme that does not involve reimbursement, was not available in the Sniiram database. Similarly, zinc sulphate, which is a pharmacy preparation, is not listed in this national database. Liver transplantation was investigated using hospital diagnoses: codes for liver transplantation in 2013 or codes for follow-up of liver transplant recipients. Existence of at least one reimbursed outpatient medical or paramedical consultation in 2013 was also investigated.

The crude prevalence rate was calculated from the number of cases identified during the study period. Regional prevalence rates (place of residence) were standardized for gender and age using the whole study population. Use of Sniiram data by the CNAMTS has been approved by decree

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