Lumanity

EPH83

Epidemiology of Wilson's Disease in Europe: A Literature Review

Authors: Rishabh Verma[1] and Amit Ahuja[1]

Affiliations: 1. Lumanity, Gurugram, India

INTRODUCTION

- Wilson's disease (WD) is a rare, autosomal recessive disorder caused by impaired copper metabolism leading to accumulation in the liver, brain and eyes[1]
- Epidemiological data in Europe are fragmented and inconsistent due to differences in diagnostic criteria, data sources, and study designs. Understanding the prevalence and incidence of WD can help inform diagnosis, care planning, and public health strategies

OBJECTIVES

The objective of this literature review is to identify and summarize published evidence on the epidemiology of WD in Europe, including estimates of prevalence and incidence reported across countries.

METHODS

- A structured literature review using Cochrane rapid review guidance[2] was conducted using predefined inclusion and exclusion criteria (Table 1) to identify relevant published articles
- MEDLINE and Embase databases were searched via Embase.com to identify English-language studies on adults with Wilson's disease from European countries. No date limits were applied to full-text manuscripts, while conference abstracts were restricted to 2020–June 2025. Studies reporting incidence, prevalence, mortality, or survival outcomes were included.
- Study eligibility was assessed by a single reviewer, with a quality check performed by a senior reviewer to ensure accuracy and consistency

RESULTS

A total of 519 studies were screened using the pre-defined study inclusion criteria. After titles and abstract screening, 30 studies underwent full-text screening. Seven studies were finally included and extracted in the review

Study characteristics

- Seven observational studies were included, conducted across France (n = 3), Germany (n = 2), the UK (n = 1) and Finland (n = 1)
- Across studies, median age ranged from 18.5[9] to 43.4[4] years. The proportion of males ranged from 45.5% (n = 15)[7] to 60% (n = 42)[4], while females ranged from 40% (n = 28)[4] to 54.5% (n = 18)[7]
- Sample sizes ranged from 33 to 2,287 patients, with study periods spanning 1998–2019

Table 1: Study characteristics

Study name	Country	Database	Sample size	Follow-up	Outcomes reported			
					Incidence	Prevalence	Mortality	Survival
Daniel-Robin 2022[3]	France	SNDS	1,520	2009–2019	✓	✓	✓	
Fang 2024[4]	Germany	WIG2 benchmark database	70	2013–2018		✓	✓	
Fang 2024[5]	France	SNDS	2287	2010–2019		✓	✓	
Poujois 2017[6]	France	Sniiram (National Health Insurance Information System database)	906	2011–2013		✓		
Sipilä 2020[7]	Finland	CRHC	33	1998–2017	✓	✓		✓
Wahler 2020[8]	Germany	Federal Statistical Office Ambulatory billing data	1,368	2005–2017 2009–2017		✓		
Wijayasiri 2021[9]	UK	NUH NHS Trust	1,794	2011–2018		✓		

Key: CRHC, Care Register for Health Care; NHS, National Health Service; NUH, Nottingham University Hospital; SNDS, Système National des Données de Santé (French national health system main database); WIG2, Wissenschaftliches Institut für Gesundheitsökonomie und Gesundheitssystemforschung; Scientific Institute for Health Economics and Health Systems Research.

Prevalence

France

- Daniel-Robin et al.[3] reported prevalence rates ranging from 1.3 per 100,000 in 2009 to 2.2 per 100,000 in 2019. Fang et al.[5] estimated a crude overall prevalence of 34.06 per million between 2010–2019, while Poujois et al.[6] reported a crude prevalence of 1.5 per 100,000 in 2013
- Fang et al.[5] reported that males had a higher prevalence than females (36.4 vs 30.6 per million). Similarly, Poujois et al.[6] reported prevalence rates of 1.65 per 100,000 in males and 1.44 per 100,000 in females
- Fang et al.[5] observed the highest crude prevalence among individuals aged 18–39 years (45.1 per million) (Figure 1). A similar pattern was noted by Poujois et al.[6], with the 20–29-year age group showing the highest prevalence at 2.41 per 100,000 (Figure 2)

Germany

- Fang et al.[4] reported an average crude annual prevalence of 20.3 per million (range: 17.8–24.4 per million) between 2013–2018. When assessed over 2-year intervals, the average prevalence was 21.9 per million (range: 20–25.2 per million). Wahler et al.[8] reported a prevalence of 1 in 60,000 in 2017
- Fang et al.[4] also reported that men were more frequently affected by WD than women, with an average prevalence of 23 per million in men compared with 17 per million in women (24 and 20 per million, respectively, for the 2-year prevalence)
- The same study found the highest prevalence in the 18–39-year age group, with a mean prevalence of 26 per million across all study years, ranging from 17.9–37.2 per million (Figure 1)

Finland

- Sipilä et al.[7] reported an overall point prevalence of 0.45/100,000 (95% CI: 0.29, 0.67) in 2017, but no more than 0.35/100,000 (95% CI: 0.21, 0.55) among native Finns

UK

- Wijayasiri et al.[9] reported an overall clinical point prevalence of 15.5/million (95% CI: 7.7, 27.7). Prevalence was 16.9 per million (95% CI: 6.2, 36.9) in men and 14.1 per million in women (95% CI: 4.6, 32.8)

Mortality

France

- Daniel-Robin et al.[3] reported a mean (SD) age of death of 57.9 (23.1) years (median: 61 [IQR: 47–75]) and a mean annual mortality rate over the 11-year study period of 2.3%
- Fang et al.[5] reported a mean age at death of 61.9 years. In 2019, the crude and World Health Organization-adjusted mortality rates were 3.2% and 2.2%, respectively. The mortality rate was the highest among patients aged 65+ (Figure 3). The mortality rate was 3.7% among men and 2.9% among women
- Fang et al.[5] also reported death proportions from 2010–2019 and observed a varied trend (Figure 4)

Germany

- Fang et al.[4] reported that mortality among patients with WD was low, with only one death in 2014 reported during the study period

Incidence

France

- Daniel-Robin et al.[3] reported an incidence rate that varied from 0.8 cases per million inhabitants per year in 2018 to 2.3 cases per million inhabitants per year in 2011

Finland

- Sipilä et al.[8] reported an annual incidence of 0.016 per 100,000 person-years (95% CI: 0.0093, 0.026)

Survival

Finland

- Sipilä et al.[8] reported that patients with WD had poorer survival during the follow-up period, with a hazard ratio of 2.92 (95% CI 1.39, 6.15; p = 0.005) compared with matched controls. There was no survival difference between men and women with WD

Figure 1: Prevalence rates by age groups in France[5] and Germany[4]

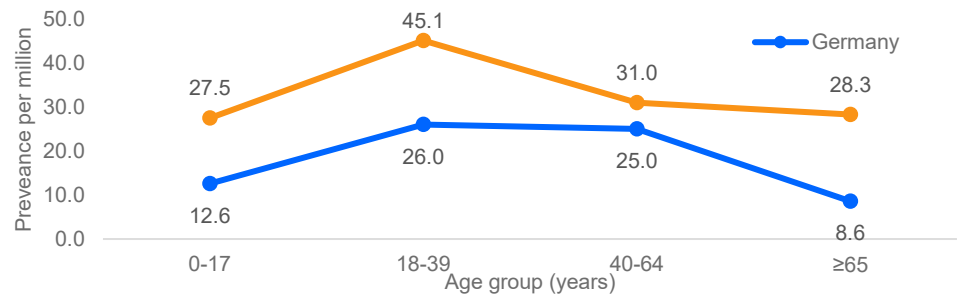


Figure 2: Prevalence rates by age group in France[6]

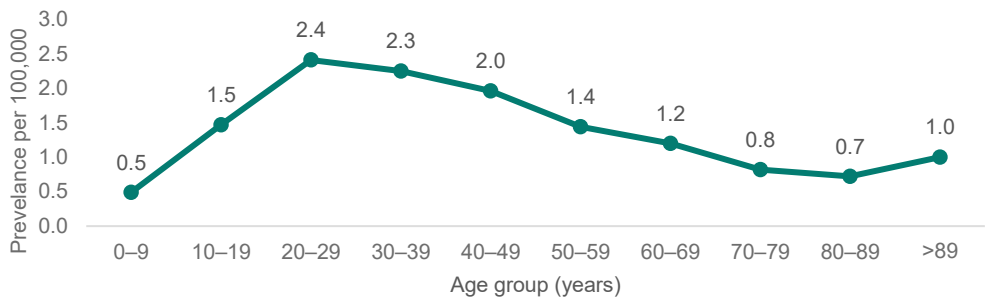


Figure 3: Mortality rate by age groups in France[5]

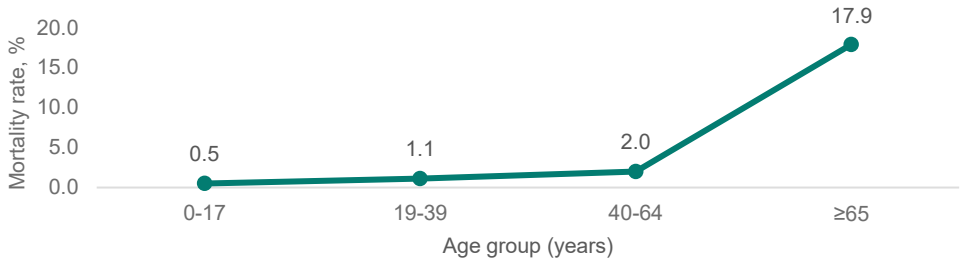
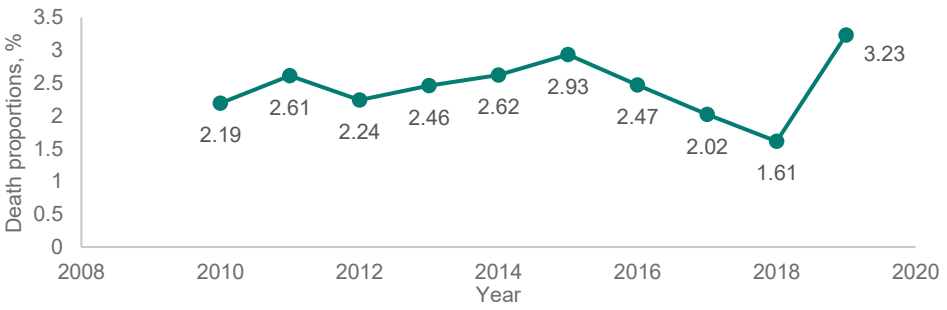


Figure 4: Death proportion in France from 2010–2019[5]



CONCLUSIONS

- Evidence on the epidemiology of WD in Europe is limited and fragmented
- Available studies suggest that regional variability is likely influenced by differences in data sources and diagnostic practices
- Younger adults and men appear to be more frequently affected
- Consistent, population-based research is needed to better understand burden and inform healthcare planning

REFERENCES

1. Bandmann O et al. Lancet Neurol. 2015 Jan;14(1):103-13. doi: 10.1016/S1474-4422(14)70190-5. PMID: 25496901; PMCID: PMC4336199. **2.** Garritty C et al. BMJ 2024 6:384:e076335. doi: 10.1136/bmj-2023-076335. PMID: 38320771. **3.** Daniel-Robin T et al. Clin Res Hepatol Gastroenterol 2022; 46: 101992. 20220703. DOI: 10.1016/j.clinre.2022.101992. **4.** Fang S et al. Orphanet J Rare Dis 2024; 19: 335. 20240911. DOI: 10.1186/s13023-024-03351-2. **5.** Fang S et al. J Inherit Metab Dis 2025; 48: e12822. 20241124. DOI: 10.1002/jimd.12822. **6.** Poujois A et al. Clin Res Hepatol Gastroenterol 2018; 42: 57–63. **7.** Sipilä JOT et al. Mov Disord 2020; 35: 2323–2327. 20200702. DOI: 10.1002/mds.28207. **8.** Wahler S at al. Value Health 2020; 23: S536. DOI: 10.1016/j.jval.2020.08.786. **9.** Wijayasiri P et al. JHEP Rep 2021; 3: 100329. 20210707. DOI: 10.1016/j.jhepr.2021.100329.

An electronic version of the poster can be viewed by scanning the QR code.

Poster presented at the 2025 ISPOR Europe; 9–12 November 2025; Glasgow, Scotland, UK.