

Population based data collection of Wilson's disease in England through a national rare disease register

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INTRODUCTION

There are approximately 3 million people in the UK affected by a rare disease. However, with few people affected by each disease, they often pose a challenge in terms of understanding prevalence and ensuring that quality evidence based care is delivered to all. Rare disease registries are a useful tool to document the natural history of disease, monitor trends, frequency and outcomes and address inequality.

MAIN TEXT

The UK strategy on Rare Diseases was published in 2013 and detailed a shared vision for improving the lives and health outcomes for affected individuals. The National Congenital Anomaly and Rare Diseases Registration Service (NCARDRS) was created in 2015 to meet this commitment.

NCARDRS is a population based register covering England and currently collects data on over a 1,000 congenital anomalies and rare diseases and is expanding to collect more rare diseases.

Wilson's disease (WD) is an inherited disorder of copper metabolism caused by mutations in the ATP7B gene which leads to excessive accumulation of copper in the body leading to complications such as cirrhosis and death.¹ WD case ascertainment is principally to improve patient outcomes by providing information on the natural history and prevalence of WD in England. It will also address healthcare equity and access to Centres of Excellence for patient across England as well as linkage to relevant Specialist Services.

METHOD

Data was collected by means of legislation granted under section 251 of the NHS 2006 Act which enables Public Health England to collect patient data, subject to strict data protection, without patient consent for the purposes of national rare disease registration. NCARDRS has access to multiple sources of patient data and identifying patients with certain conditions requires the linkage of different data sources. Ideally, all cases of WD would be identified by clinicians throughout National Health Service (NHS) hospitals in England and reported to NCARDRS to avoid patients from being missed. However, responses from clinicians were limited as it demands time from busy clinicians using patient records that are not always available.

The use of routinely collected data, such as Hospital Episode Statistics (HES) also fails to provide a true data set due to limitations of ICD 10 coding. We therefore cross referenced a variety of health data resources available to NCARDRS including:

- Primary (community) prescribing data
- Molecular results from Specialist Genetics labs
- Supra-regional Assay (SAS) trace element labs
- Office of National Statistics (ONS) mortality data
- Case confirmation by clinicians in hospitals across NHS England
- Cohort Research On Wilson's Disease: Genetic Determinants and Biomarker Discovery for Neurological Involvement (Crowd Study)

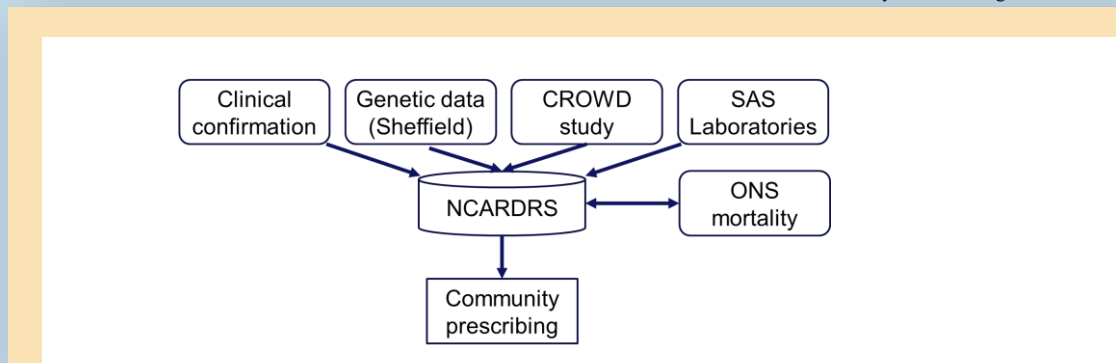


Figure 1. Variety of data sources used to identify cases of WD by NCARDRS

RESULTS

Using current data sources, we have been able to case confirm over 400 cases of WD in England to date. These include cases reported by 26 Trusts across NHS England as well as 10% identified through other sources such as primary prescription data.

CONCLUSIONS

Rare disease registries allow us to understand the epidemiology of conditions that often have significant health implications for those affected. NCARDRS was created in 2015 and has recently expanded to include Wilson's disease with the aim of determining prevalence and natural history to ultimately improve patient outcomes. Close collaboration with patient groups and clinical experts as part of a WD Special Interest Group has allowed us to reach a wider audience.

We envision a future that will include prospective registration at diagnosis through multiple sources to ensure high ascertainment as well as using the registry to advocate for pioneering research to improve outcomes

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REFERENCES

1. Coffey et al, A genetic study of Wilson's disease in the United Kingdom, *Brain*, 2013 May;136(Pt 5):1476-87

DISCLOSURES

There are no disclosures from the authors.