Wilson's disease registry in Hungary: a registry protocol

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Introduction

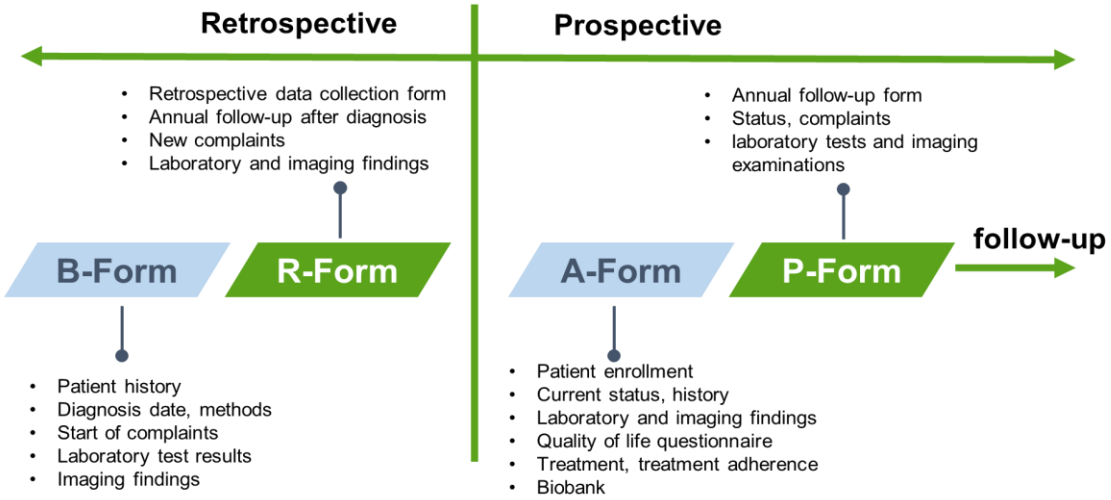
Wilson's disease is a rare, genetic disorder in which defective biliary excretion of copper leads to its accumulation, particularly in the liver and the central nervous system. A significant variance can be observed in the patterns of clinical presentation, even in individuals carrying the same mutation. There are little data on long-term outcomes including survival from cohort studies.

The registry aims to investigate the epidemiology, disease course, treatment modalities, and predictive factors of disease progression in Wilson's disease.

Disclosure: Nothing to disclose

Methods

The registry for Wilson's disease received the ethical and biobank permission in 2018 (TUKEB-11862-9/2018/EKU). This multicenter, prospective, national registry will initially involve 4 centers in Hungary. Participation is voluntary and based on written consent. Any patient diagnosed with Wilson's disease will be included upon presentation.



Since the registry is an observational study, all intervention will be performed only if justified with medical indication. Details from medical history, status, complaints, laboratory studies, imaging, and therapy will be prospectively collected on enrollment. Lifelong annual follow-up with data recording is planned for every patient. The registry is open for all national centers which can provide data on patients with Wilson's disease. In the future, we plan to extend the authority of the registry to other countries.

Conclusions

Besides providing valuable data and research material, the registry may help to standardize the management of patients with Wilson's disease and facilitates the establishment of a network for potential research cooperation first in Hungary, then in Central and Eastern Europe.