Editorial for focused issue "Wilson Disease: From Genetics to Management of Disease"

Wilson disease: current knowledge, challenges and perspectives

Wilson disease was previously called hepatolenticular degeneration. It is a rare inherited disorder preventing the body from removing copper via bile. Accordingly, copper builds up in liver and spills over to other organs, i.e., the basal ganglia of the brain. The increasing copper burden is believed to provoke reactive oxygen species formation and mitochondrial disintegration triggering cell death. Although the symptoms vary among and within families, the most common manifestation of Wilson disease is progressive liver damage combined with a wide spectrum of neurological and psychiatric disturbances. Presently, medical therapy is based on the lifelong treatment with copper chelating agents or zinc. Dietary restriction is of minor importance. Several new therapy options are presently experimentally or clinically tested. During the last years, guidelines for screening of patients with unexplained liver, neurological, or psychiatric abnormalities have been published. Patients who are recognized early to have Wilson disease and receive adequate treatment have a good long-term prognosis. In case of liver failure and decompensated cirrhosis, liver transplantation is an option. However, several questions remain unresolved including genotype-phenotype relationship in disease manifestation, impact of epigenetic factors, diagnostic challenges, establishment of individualized therapies, combination therapy and best medication options of neurological or psychiatric disorders.

In this focused issue "*Wilson Disease*: *From Genetics to Management of Disease*", leading experts working in the field of Wilson disease have summarized recent findings and advances in our understanding of this copper overload disease in regard to experimental and clinical aspects. The present knowledge on genetics and pathophysiology in relation to diagnostic and therapeutic procedures is our special interest. Challenges in diagnosis, anti-copper therapies, and clinical practice guidelines will be discussed. This compilation is an important source of information for scientists, clinicians and patients (*Figure 1*).

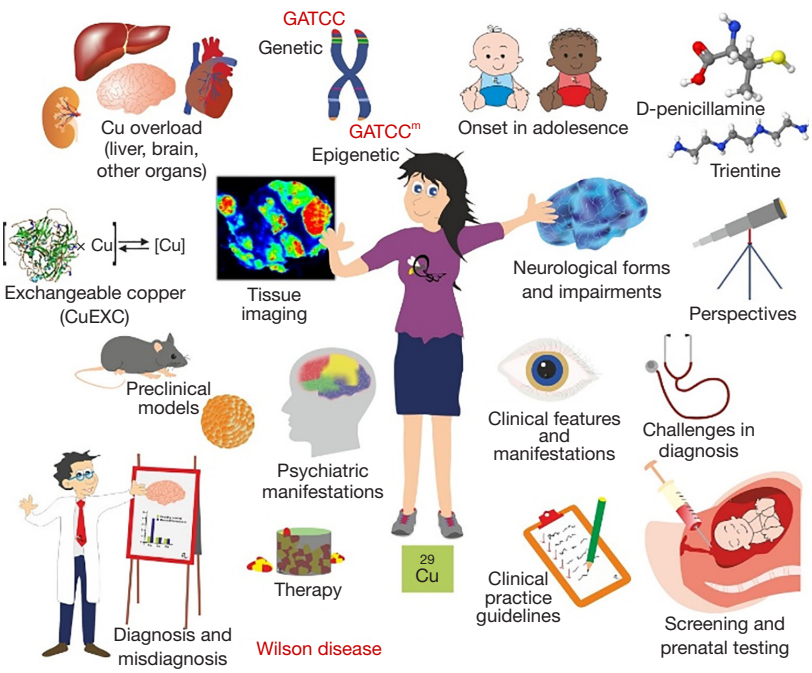

Figure 1 Topics covered in this special issue. This special issue contains expert reviews on the genetics and epigenetics of Wilson disease, the pathogenesis, diagnosis, clinical management, complications, preclinical models, and therapies on this copper overload disease. The different contributions discuss highlights in basic and clinical research and summarize up-to-date facts and perspectives that are of current interest in basic research and clinical presentation of Wilson disease.

We are grateful to all authors that have contributed excellent overviews to this special issue. In addition, we cordially thank the Editors-in-Chief of *Annals of Translational Medicine*, Drs. Xuehao Wang, Jianxing He, and Rafael Rosell, for the opportunity to serve as Guest Editors for such an appealing collection of articles. Moreover, we would like to acknowledge the kind support of the Editorial office team from *Annals of Translational Medicine*. Special gratitude goes to the Senior and Science Editors Nikki Ling, Nicole J. Li, and Vicky Wong who were extremely important in helping us to arrange a strict review process for this compilation and by assisting us to enable a reliable correspondence with the authors. Last but not least, we would like to emphasize the excellent and highly efficient work of the reviewers, who were eminent important for the overall quality of this special issue.

Acknowledgements

The authors would like to thank Sabine Weiskirchen for drafting of *Figure 1*.

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doi: 10.21037/atm.2018.12.06
Conflicts of Interest: The authors have no conflicts of interest to declare.
View this article at: http://dx.doi.org/10.21037/atm.2018.12.06

Cite this article as: Weiskirchen R, Stremmel W. Editorial for focused issue "Wilson Disease: From Genetics to Management of Disease". Ann Transl Med 2019;7(Suppl 2):S55. doi: 10.21037/atm.2018.12.06

