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Wolfram Syndrome 1: From Genetics to Therapy

Guest Editor:

Dr. Luciana Rigoli
Department of Human Pathology in Adulthood and Childhood "G. Barresi", University Hospital "G. Martino", Via C. Valeria 1, 98125 Messina, Italy

Deadline for manuscript submissions:
closed (15 September 2023)

Message from the Guest Editor

Dear Colleagues,

Wolfram syndrome 1 (WS1) is a rare neurodegenerative disease transmitted in an autosomal recessive mode. The main clinical features are diabetes insipidus (DI), diabetes mellitus (DM), optic atrophy (OA), and deafness (D), hence the acronym DIDMOAD. It is frequently complicated by other symptoms, such as urinary tract, endocrinological, psychiatric, and neurological abnormalities. Early-onset non-autoimmune insulin-dependent DM and bilateral OA are key clinical criteria for the diagnosis of WS1. WS1 is caused by mutations in the WFS1 gene located on chromosome 4p16 that encodes a transmembrane protein named wolframin. Wolframin plays a key role in the regulation of ER calcium homeostasis and, therefore, in cellular apoptosis. More than 200 mutations are responsible for WS1.

This special issue focuses on the molecular pathology of wolfram syndrome 1 and explores potential treatment options from genetics. For any of these topics, this Special Issue is open to contributions, both in the form of reviews and research articles.

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Editor-in-Chief

Prof. Dr. Maurizio Battino
Department of Odontostomatologic and Specialized Clinical Sciences, Sez-Biochimica, Faculty of Medicine, Università Politecnica delle Marche, Via Ranieri 65, 60100 Ancona, Italy

Message from the Editor-in-Chief

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International Journal of Molecular Sciences Editorial Office
MDPI, Grosspeteranlage 5
4052 Basel, Switzerland

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