Special Issue

Wolfram Syndrome in Pediatric Age

Message from the Guest Editor

Wolfram syndrome 1 (WS1; OMIM 222300) is a rare, autosomal recessive, neurodegenerative, and progressive disease, also known by the acronym DIDMOAD (diabetes insipidus DI, diabetes mellitus DM, optic atrophy OA, and deafness D). WS1 is an autosomal-recessive disorder usually diagnosed in childhood when non-autoimmune, insulin-dependent diabetes is associated with optic atrophy. Additional clinical manifestations include ureterohydronephrosis, neuropsychiatric and endocrinological impairment, and cataract. WS1 prevalence in the general population has been reported to be from 1/770,000 individuals to 1/54,478 in different ethnic groups. WS1 is caused by mutations in the WFS1 gene located on 4p16.1 which encodes wolframin, an 890-amino-acid glycoprotein which is involved in the regulation of endoplasmic reticulum (ER) stress responses.

Guest Editor

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