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Wolfram Syndrome: A rare multiorgan disease

Wolfram Syndrome (WS) is a rare neurodegenerative disease with autosomal recessive inheritance and characterized by diabetes mellitus and optic atrophy, followed by diabetes insipidus, deafness and several endocrine and neurological dysfunctions, affecting the central nervous system and including anosmia, ataxia, seizures, nystagmus, gaze palsies, dysarthria, dysphagia, psychiatric disturbances, cognitive impairment, neurogenic bladder, central apnoea, neurogenic upper airway collapse myoclonus. The acronym DIDMOAD (Diabetes Insipidus, Diabetes Mellitus, Optic Atrophy Deafness) includes the main clinical characteristics of the syndrome. WS is a rare disease with an estimated prevalence of 1/550.000 of children, and with a carrier frequency of 1/354, and is caused by biallelic mutations of the WFS1 gene located on chromosome 4p16.1 and encoding Wolframin, a transmembrane glycoprotein in the endoplasmic reticulum (ER). WFS1 is localized also in the brain and ER stress-mediated cell death could be responsible of neurodegeneration. Mortality is about 65% before age 35, mainly due to respiratory failure or dysphagia secondary to brainstem impairment. The efforts for determining WS natural history have clarified the order of onset of the various clinical symptoms. Noteworthy the molecular complexity of the syndrome, the wide spectrum of associated diseases and the small sample size of patients evaluated made difficult to establish a correlation between genotype and phenotype. Brain atrophy is described in the late stage of the disease, otherwise little is known either about at what stage of the disease neurological abnormalities appear, the role of WFS1 mutations during neurodevelopment, the correlation between neurological phenotype and genotype.

Biography

Giuseppe d'Annunzio has completed his degree in Medicine in 1985, Post-graduation in Preventive Pediatric and Puericulture in 1989 and Post-graduation in Endocrinology at the University of Pavia in 1994. He is the responsible for Regional Center for Pediatric Diabetes, Giannina Gaslini Institute, Genoa. At present he is author/co-author of 104 papers in reputed journals (H index 17), has been serving as reviewer of several journals, and as an Editorial Board Member of repute.

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