

Common Manifestations and Natural History of Wolfram syndrome

The common manifestations of Wolfram syndrome include: diabetes mellitus, optic nerve atrophy, central diabetes insipidus, sensorineural deafness, urinary tract problems and progressive neurologic difficulties. Diabetes mellitus is typically the first manifestation, usually diagnosed around age 6. Optic nerve atrophy, marked by loss of color vision and peripheral vision, follows around age 11. Central diabetes insipidus is another common manifestation, affecting approximately 70 percent of Wolfram. Around 65 percent of patients develop sensorineural deafness that can range in severity from deafness beginning at birth to mild hearing loss beginning in adolescence that worsens over time. Urinary tract problems are another major clinical challenge for Wolfram syndrome patients affecting 60 to 90 percent of this population. These problems include obstruction of the ducts between the kidneys and bladder, high-capacity atonal bladder, disrupted urination, bladder sphincter dyssynergia, and difficulty controlling urine flow. About 60 percent of patients with Wolfram syndrome develop neurological manifestations, most commonly presenting as problems with balance and coordination (ataxia) beginning in early adulthood. Brain stem atrophy is also a prominent feature that often results in death secondary to central apnea. Clinical manifestations related to mood disorder and autonomic dysfunction are commonly seen.

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