

A Case Report of Wolfram Syndrome due to a novel homozygous mutation in *WFS1* gene

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Introduction: Wolfram Syndrome (WFS: OMIM 222300), also known as DIDMOAD

(Diabetes Mellitus, Optic Atrophy and Deafness) is an autosomal recessive, progressive, neurologic, and endocrinologic, degenerative disorder caused by mutation in the *WFS1* Gene.

This report presents a case with a new defined mutation in Wolfram Syndrome.

Case Presentation:

14 years old male patient was diagnosed with non-autoimmune type I diabetes at the age of 5 and insulin treatment was applied to him. He has been diagnosed optic nerve degeneration at 7 years old and diagnosed sensory neural hearing loss at the age of 9 and implanted cochlear implants to him. His parents were not relative. Physical examination revealed that body weight 52,5 kg (-0,95 SDS), height 157,5cm (0,95 SDS), testicular volume 5/5 cc, pubic and axillary hair were consistent with stage 2. There were no clinical and lab findings about diabetes insipidus. Neurological examination and cranial imaging were normal. Genetic testing for *WFS1* gene mutation was performed and a new homozygous mutation in *WFS1* gene were identified in the genetic analysis (c.1536-1549dupCTATCTCTTCTTC).

Conclusion:

Non- autoimmune diabetes mellitus is the first and the most frequently seen symptom of Wolfram Syndrome. Optic atrophy, and deafness generally appears in the first decade of patients. Diabetes insipidus, renal and neurologic signs may appear in the second 10 years. Patients should be monitored multidisciplinary. Genetic studies will provide early diagnosis and genetic counseling for families.