

9-year old boy with Wolfram syndrome - case report

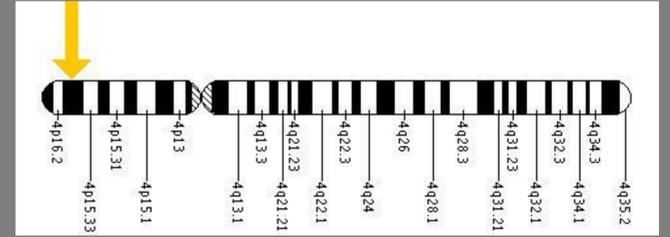


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Introduction

Wolfram syndrome is a rare progressive genetic neurodegenerative disorder connected with diabetes mellitus, diabetes insipidus, optic atrophy, deafness, neurologic and endocrine abnormalities. Wolfram syndrome is inherited in autosomal recessive manner- due to mutation of the WFS1 gene which is located on chromosome 4.

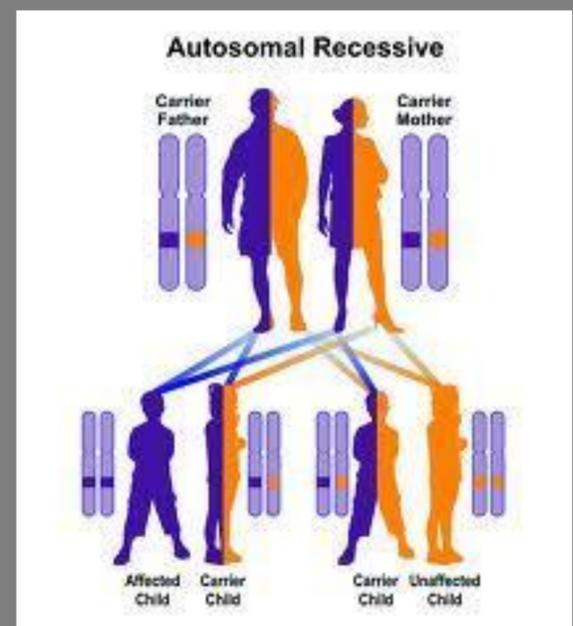


Location of mutations leading to Wolfram Syndrome

Case history

9-year old boy, diagnosed with diabetes mellitus at the age of 5,5 years, was admitted to hospital for further investigation of optic atrophy and polydipsia with polyuria while euglycaemic. Patient did not necessitate pharmacological treatment (unless having increased insulin requirement- such as during infection) for diabetes mellitus, only followed diabetic diet. Blood samples were taken for the genetic tests on appointment to outpatient clinic-as presented symptoms grew suspicion for Wolfram syndrome.

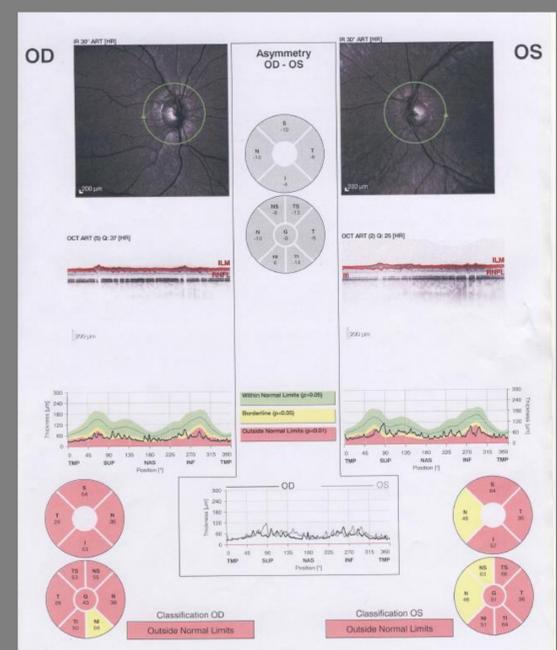
Physical examination revealed correct psychomotor evaluation, lateral nystagmus and malocclusion. Laboratory tests showed anaemia and thrombocytopenia. Hormonal tests excluded central diabetes insipidus. Abdominal ultrasound and MRI scan of central nervous system showed no abnormalities. Laryngological examination demonstrated sensorineural hearing deficits of high-frequency in left ear and mixed deficit in right ear.



Inheritance pattern of Wolfram Syndrome

Results

Ophthalmic examination and Optical Coherence Tomography of the optic nerve discs revealed optic nerve discs thinning, global thinning of bundles of neural fibres-confirming the diagnosis of optic nerve neuropathic atrophy. During hospitalisation results of genetic tests were obtained and the diagnosis of Wolfram syndrome was acknowledged.



Location of mutations leading to Wolfram Syndrome

Conclusion

Wolfram syndrome may be a rare cause of diabetes mellitus, and patients with such diagnosis require multispecialist care and genetic counselling.