Editorial

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Editorial on Wolfram Syndrome

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Editorial

Wolfram syndrome or DIDMOAD syndrome a rare genetic disorder that is typically associated with childhood-onset insulin-dependent diabetes mellitus and progressive optic atrophy. People with Wolfram syndrome often also have pituitary gland dysfunction that results in the excretion of excessive amounts of urine (diabetes insipidus), hearing loss caused by changes in the inner ear. There are 2 types of WS may be distinguished: type 1 (WS1) and type 2 (WS2).

This syndrome is caused by the mutations in the *WFS1* gene that is responsible for more than 90 percent of Wolfram syndrome type 1 cases. *WFS1* gene provides instructions for producing a protein called wolframin and regulate the amount of calcium in cells. A proper calcium balance is required for many different cellular functions i.e. protein processing, cell-to-cell communication and the contraction of muscles. *CISD2* encodes the ERIS protein and a certain mutation in the *CISD2* gene was found to cause Wolfram syndrome type 2. ERIS protein has roles in roles in glucose homeostasis regulation, insulin sensitivity for calcium homeostasis and autophagy. Wolfram syndrome is inherited in an autosomal recessive pattern. The estimated occurrence of Wolfram syndrome type 1 is 1 in 500,000 people worldwide [1].

The primary symptoms of Wolfram syndrome includes diabetes insipidus, childhood-onset diabetes mellitus, gradual loss of vision due to optic atrophy and deafness. Some others symptoms include: Urinary tract abnormalities, Neurological symptoms, Psychiatric and behavioral problems, gastrointestinal problems and Disordered sleep etc. [2].

A diagnosis is confirmed by genetic screening and based on the presence of characteristic signs and symptoms. The clinical criteria for WS diagnosis are juvenile-onset diabetes mellitus and bilateral optical atrophy, family history of Wolfram syndrome or diabetes mellitus and sensorineural deafness [3].

Management and treatment of Wolfram syndrome is supportive based on the signs and symptoms. It includes an annual screening for diabetes mellitus, vision, sensorineural deafness, urodynamic testing, nephropathy and daily insulin injections and a controlled diet to treat diabetes mellitus. People with hearing loss may benefit from hearing aids or cochlear implantation. Treatment of diabetes insipidus, apnea and urinary disorders are needed.

References

1 https://ghr.nlm.nih.gov/condition/wolfram-syndrome

2 https://rarediseases.info.nih.gov/diseases/7898/wolfram-syndrome

3 https://rarediseases.org/rare-diseases/wolfram-syndrome/