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Assessment Of Neuroradiological Features Of Wolfram Syndrome

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Abstract
The WOLFRAM syndrome is a rare hereditary neurodegenerative disorder described for the first time in 1938 as the coexistence of a diabetes mellitus and optic atrophy to which join frequently a diabetes insipidus and a bilateral deafness. Here we report cranial MRI findings in a 20 year old patient who presented with diabetes mellitus, decreased visual loss since 3 years with progressive loss of hearing since 6 months. We reviewed the brain MR images to determine the frequency and characteristics of common neuroradiologic findings. Imaging findings include absence of the normal high signal of the neurohypophysis, atrophy of visual pathways, the brainstem, cerebellum and cerebral cortex and abnormal signal changes in pons.
Keywords: *Atrophy, brainstem, hypophysis, optic nerves.*

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I. Introduction

WOLFRAM syndrome is a rare neurodegenerative disorder characterized by the coexistence of diabetes mellitus, optic atrophy and deafness. DIDMOAD is an acronym made from the disease components i.e. diabetes insipidus, diabetes mellitus, optic atrophy, and deafness. It is also associated with a wide variety of abnormalities of the central nervous system, urinary tract and endocrine glands. This cluster of findings was first described in medical literature in 1938 by clinicians, one of whom was named Wolfram, in sets of siblings. It is caused by a mutation in the WFS1 gene that encodes wolframin, a transmembrane protein of pancreatic β cells.1 The life expectancy of patients diagnosed with this syndrome is about 30 years.

II. Case Report

A 20-year-old man had diabetes insipidus, diabetes mellitus, and visual loss since 3 years with progressive loss of hearing since 6 months. He was clinically diagnosed with Wolfram syndrome on the basis of the typical clinical features and familial history of diabetes mellitus. The man was admitted to our hospital because of consciousness disturbance. Laboratory examinations showed diabetic ketoacidosis. He was stuporous and opened his eyes only to painful stimuli. After the patient recovered from diabetic ketoacidosis, neurologic examinations revealed complete visual loss and mild sensory hearing loss.

On imaging, **non-contrast axial MRI Brain** showed diffuse bilateral cerebral & cerebellar atrophy (predominantly cerebellar and vermis) as evidenced by prominent sulcal and folial spaces (figure 1). **On T1WI sagittal images**, there is diffuse atrophy of brainstem (including midbrain, pons and medulla) and cerebellar vermis (figure 2). Also, T1 bright spot of posterior hypophysis is not appreciated however anterior hypophysis appears normal in morphology.

On T2WI, abnormal signal changes (hyperintense signal) involving tegmentum of midbrain and anterior pons (figure 4). **On CISS 3D sequences,** diffuse atrophy of bilateral optic nerves (measuring ~1.5 -2 mm) with resultant prominent CSF sleeve around (figure 3). Rest of the brain parenchyma shows normal gray/white matter differentiation. The ventricles, sulcal and basal cisterns are dilated. Cranio-vertebral and cervico-medullary junctions are normal.

III. Discussion

Wolfram syndrome (WFS) is a rare hereditary neurodegenerative disorder also known as DIDMOAD (diabetes insipidus, diabetes mellitus, optic atrophy, and deafness). It is characterized by the coexistence of diabetes mellitus, diabetes insipidus, optic atrophy and deafness. Several other manifestations are frequently associated: neurological, urologic and endocrinous abnormalities as well as growth troubles. Only diabetes mellitus and optic atrophy are compulsory for the diagnosis. In addition to the classical features, including absence of the normal high signal of the neurohypophysis, atrophy of visual pathways, the brainstem, cerebellum and cerebral cortex, we observed abnormal signal changes in tegmentum and pons on T2 weighted images.

IV. Conclusion

Patients with Wolfram syndrome present characteristic neuroradiologic findings that involve the posterior pituitary gland, optic nerves and chiasm, cerebral white matter, brain stem, and cerebellum. These abnormal findings appear at an early age and tend to increase in frequency with time. Neuroradiologists should be aware of these findings when reading MR imaging studies of patients with Wolfram syndrome. Future research could pursue the diagnostic and prognostic value of these signs when combined with quantitative neuroimaging data and the pathophysiologic processes underlying these signs. In conclusion, WFS is a devastating disease for the patients and their families. More information about WFS will lead to a better understanding of this disease and hopefully to improvement in means of its prevention and treatment.

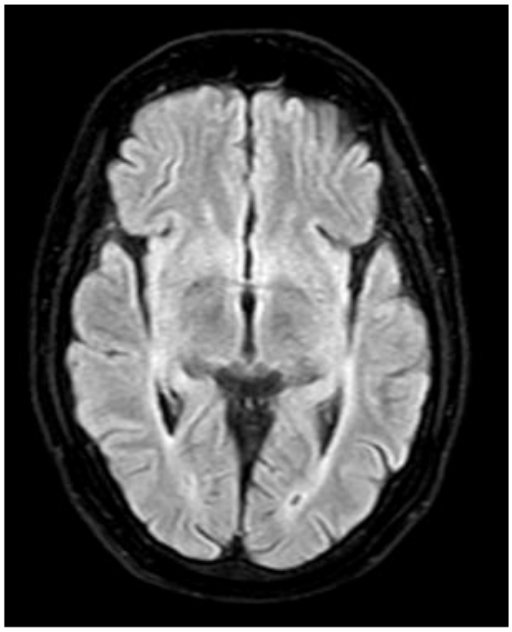

Figure 1: Axial non-contrast MRI Brain showing diffuse bilateral cerebral and cerebellar atrophy.

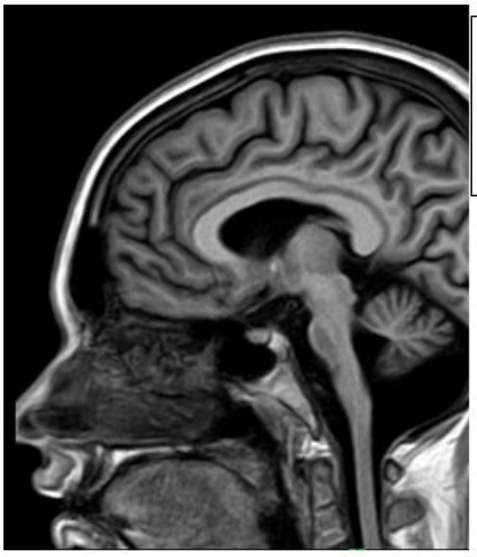

Figure 2: T1 sagittal images showing diffuse atrophy of brainstem (including midbrain, pons and medulla) and cerebellar vermis.

Absence of T1 bright spot of posterior hypophysis.

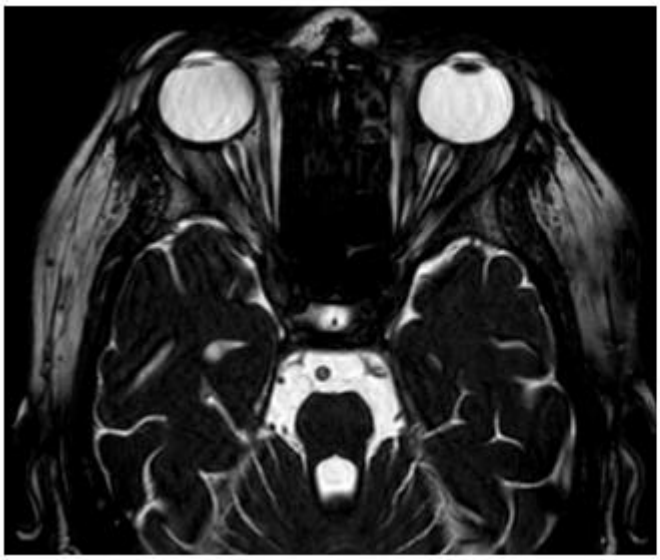

Figure 3: CISS 3D image showing diffuse atrophy of bilateral optic nerves with resultant prominent CSF sleeve around.

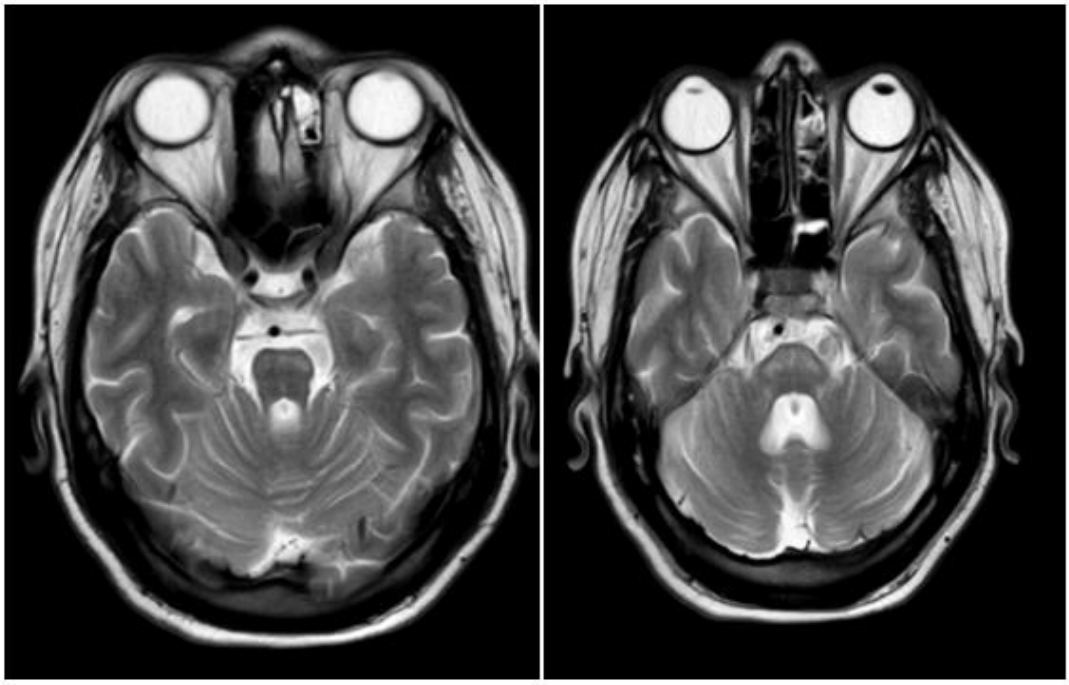

Figure 4- Abnormal signal changes involving tegmentum of midbrain and anterior pons