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Title: There's More Than Meets the Eye: Wolfram Syndrome

Wolfram syndrome (WS) is a rare neurodegenerative and genetic disorder, also known by the synonym DIDMOAD, which stands for diabetes insipidus (DI), childhood-onset diabetes mellitus (DM), optic atrophy (OA), and deafness (D).

We present a case of a 38 year old year-old who was diagnosed as type one diabetes patient , and was treated along those lines all along, had indications of diabetic retinopathy deafness, secondary amenorrhea and vitamin D deficiency She is suspected to be affected with mitochondrial diabetes or drug induced hyperprolactinemia and was evaluated for pathogenic variations in Gene ABCC8, APPL1, CAV1, GATA6, INS,INSR , BLK, CEL, FOXP3, GCK,HADH KLF11 Gene testing revealed , negative for mitochondrial inheritance but ,WFS1 (+) on exon 8(c.1530c)A variant, with homozygosity , with disease suggestion of Wolfram syndrome-1 , autosomal recessive . Classified as pathogenic (PVS1, PM2, PP3). After workup, a diagnosis WFS was made. Once the diagnosis was reached, treatment was subcutaneous insulin and nasal desmopressin it’s the patient's symptoms. In juvenile diabetic patients presenting with new onset or worsening polyuria and polydipsia, the possibility of WS should be considered. Early diagnosis and initiation of appropriate management leads to improved outcomes and the quality of life.

Keywords: DIDMOAD; Deafness; Diabetes type I; Optic atrophy; Wolfram syndrome.