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Title

The combination of atypical diabetes with atypical optic atrophy, avoid missing the diagnosis of wolfram syndrome.

Priority 1 (Research Category)

Practice management and organization

Presenters

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Abstract

Abstract: Objective: Our goal is to create awareness of atypical diabetes with atypical optic atrophy in children and adolescents through multidisciplinary approach leading to early diagnosis of Wolfram Syndrome and improving quality of life. Presentation: A 5-year-old Pakistani decent American born male in parents with consanguineous marriage initially presented to PCP with complaint of decrease growth, weight loss, polydipsia, polyphagia, and urinary incontinence. Work up done showed negative antibodies (GAD 65, IA2, Insulin AB) but A1c of 11. Patient diagnosed with insulin dependent Non autoimmune atypical DM type 1. Patient was seen by ophthalmology for baseline eye exam in new onset childhood DM type 1 in 6 months. Patient got diagnosed with optic atrophy (based on exam 20/80 in both eyes) at the age of 6 years and recommended to obtain brain MRI to rule out intra cranial tumor or mass as cause of atrophy which came back negative and correction glasses was advised. At the age of 6.5 years of age, Patient then presented with worsening school grades with difficulty in vision and hearing. Patient underwent 2nd eye exam showed: Atypical Optic Atrophy. Vision reduced to 20/200 in both eyes. Patient underwent genetic disorders screening. Genetics: At the age of 7, sequencing analysis and deletion/duplication testing showed positive for WFS1 gene. WFS1 gene is involved in rare autosomal recessive genetic disorder called wolfram syndrome coined also DIDMOAD (diabetes insipidus, diabetes mellitus, optic atrophy, and deafness). ENT/Audiology: diagnosed the patient with bilateral Sensi neural hearing loss and started on hearing aids. Endocrinology: Patient underwent further testing and was found to have GH def and negative DI. Unique about our case: Most of the studies reported median age of onset of optic atrophy is 11 years with other studies ranging from age 8-13 years. Our patient was diagnosed with optic atrophy at the age of 6. The earliest optic atrophy in wolfram syndrome has ever been reported. Central DI was absent in our patient with WS1, although most cases of WS2 have absent DI. The variant c 1028 T>C in exon 8 pathogenic in our study has not been studied before in this syndrome. Conclusion: Wolfram syndrome is a rare genetic disorder with neurodegenerative features and multi organ failure. Our study showed the vital requirement of multidisciplinary healthcare approach for identification of Wolfram syndrome to prevent delay in care.