Visual disturbance in Diabetes Mellitus; don’t be blind to alternatives to retinopathy

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Learning points
• Ophthalmic pathologies are well-recognised complications of Diabetes Mellitus
• Retinopathy screening is indicated at all ages²
• Alternative ophthalmic/non-ophthalmic aetiologies must be considered
• Diabetes Mellitus with optic atrophy should be investigated for Wolfram Syndrome
• Additional diagnoses cause cumulative stresses on the child and family, which can hinder disease management and treatment compliance

Case Summary
• Diabetes Mellitus diagnosed at 4 years old. Vision problems began at age 8
• No retinopathy. Impaired colour vision and significant bilateral optic disc pallor
• Normal neurological examination and cranial MRI
• No mitochondrial DNA deletion to suggest Leber’s optic atrophy
• Parental consanguinity suggests a possible genetic aetiology
• Child is homozygous for WFS1 gene mutation (type 1 Wolfram Syndrome)

• Use of an insulin pump provides additional benefits given her vision problems
• Diabetes control had been poor, which hindered surveillance for the onset of Diabetes Insipidus
• Developed Diabetes Insipidus at 13 years old
• Hearing currently unaffected and renal ultrasound normal

• In view of the family history, when their 2 year old sibling developed Diabetes Mellitus aged 2 they were tested for the WFS1 mutation, which was present. Wolfram Syndrome was diagnosed before the onset of optic atrophy in this child

Wolfram Syndrome (type 1)¹
• Autosomal recessive
• >90% due to mutation in WFS1 gene
• Estimated worldwide prevalence 1 in 500,000
• First presentation is usually Diabetes Mellitus
• Optic atrophy
• Diabetes Insipidus, sensorinueral deafness, urinary tract pathology, hypogonadism (in males)
• Neurological and psychiatric disorders, commonly ataxia in early adulthood

Ophthalmic pathologies in patients with Diabetes Mellitus
- Corneal erosions
- Delayed wound healing
- Choroidopathy
- Retinopathy
- Neuropathy
- Corneal ulcers
- Non-diabetic eye disease
- Corneal oedema

Natural history of Wolfram Syndrome. The proportion of patients (density) for each clinical feature at onset age, measured as a nonparametric probability density distribution: DE, deceased; DI, diabetes insipidus; DM, diabetes mellitus; HD, hearing defects; ND, neurological, psychiatric, and developmental defects; OA, optic atrophy; UD, urological or renal defects

Ocular pathology photos: Primary optic atrophy: http://tedmontgomery.com/the_eye/eyephotos/index.html [website]