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RESEARCH ARTICLE

WOLFRAM SYNDROME: A CASE REPORT

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Abstract

Wolfram Syndrome is a severe genetic disease, including diabetes, bilateral optic atrophy, and other systemic disorders. We present the case of a diabetic patient who presented with decreased visual acuity and was diagnosed with Wolfram syndrome.

Introduction:-

Wolfram Syndrome is a seriousgeneticdiseasecombiningdiabetes, bilateralprimaryopticatrophy and sensorineuraldeafness, whichbeginsbetween the first and second decade of life. (1).

Observation:-

The patient was 25 yearsold and had a history of grade 1 diabetes. Shepresentedwith a progressive decrease in visualacuitysincechildhood. Clinicalexaminationrevealedreducedvisualacuityat light perception, oculartoneat 28 mmhg on the right and 30 mmhg on the left. In the right eye, the anterior segment was normal. In the lefteye, the examinationrevealedexotropia, pupillarydeficit, areflexic semi-mydriasis, posteriorsynechiaat 4 o'clock, and a subtotal cataract. Examination of the posterior segment revealedbilateralopticatrophy. MRI revealed a significantdecrease in optic nerve signal, and color vision wasimpaired. ENT examinationrevealedsensorineuralhearingloss. The rest of the generalexaminationruled out otherabnormalities. Giventhisclinicalpicture, the diagnosis of wolfram syndrome was made. Geneticanalysisrevealed a mutation in the wFS1 gene. Treatmentconsisted of diabetes monitoring, hypotonizingtherapy, annual screening for othersystemicabnormalities, and psychological support.

Discussion:-

Wolfram syndrome, originallycalled DIDMOAD (2), is an oto-optico-diabetic syndrome with an estimatedprevalence of 1/700,000(3). There are twoclinicalforms, transmittedautosomalrecessively (3). Diagnosisisbased on the presence of two major criteria, or one major and twominorcriteria. Major criteriaincludeinsulin-dependentdiabetes and opticatrophy, whileminorcriteriainclude minimal to moderatedeafness and diabetesinsipidus (4). Lessfrequentocularabnormalitiesincludeabnormalpupillary reflexes, nystagmus, cataract, pigmentarymaculopathy, retinopathy and glaucoma. Othersystemic manifestations have been described, includingneurologicaldisorders, psychiatricdisorders and dementia (3). There is no treatmentavailable to limit the progression of opticneuropathy. Gene therapyiscurrentlyunderstudy (4).

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Conclusion:-

Wolfram syndrome is a geneticdiseasethatcan affect severalorgans, including the eye. Screening issystematic in the presence of afamily case. It is a seriousdiseaserequiring long-termfollow-up and symptomatictreatment, not to mention psychological support for patients and theirfamilies.

Fig 1:- Photographie de la patiente montrant l'exotropie et la cataracte de l'œil gauche.

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