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Gyrus atrophy of the choroid and retina. A case presentation

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Gyrus atrophy is a rare autosomal recessive hereditary disease secondary to a mutation of the OAT gene on chromosome 10 which results in a deficiency of the mitochondrial enzyme ornithine aminotransferase that causes a 20-fold increase in serum concentrations of the amino acid ornithine. The disease is characterized by atrophic choroid and retinal patches that begin in the peripheral retina in the first decade and later spread centrally to the macular area, causing cystic changes and posterior subcapsular cataracts.

The clinical picture is characterized by night blindness, constriction of the visual field and finally decreased central vision and blindness. In this article, we present the case of a 53-year-old woman with no significant hereditary antecedents who presented a decrease in visual acuity after several months of evolution.

Introduction. Gyrus atrophy is an autosomal recessive hereditary disease secondary to a mutation of the OAT gene on chromosome 10 which results in the deficiency of the mitochondrial enzyme ornithine aminotransferase that catalyzes the transamination of L-ornithine to 5-glutamate semialdehyde using pyridoxine as a cofactor; with subsequent conversion to alpha-ketoglutarate and glutamate. Therefore, enzyme deficiency causes a 20-fold increase in serum concentrations of the amino acid ornithine. It is considered a rare disease, worldwide incidence is 1: 1,500,000 inhabitants, frequently beginning during late childhood and rarely after the fourth decade of life. [1].

It is characterized by atrophic chorioretinal patches that begin in the peripheral retina in the first decade and later spread centrally to the macular area, causing cystic changes and posterior subcapsular cataracts.

The clinical picture is characterized initially by night blindness and myopia during the first decade of life, later visual field constriction and finally decreased central vision and blindness between the fourth and sixth decade of life.

Diagnosis is made by compatible medical history, suggestive ophthalmoscopy (areas of choroid and retinal atrophy that progressively fuse and have pigmented edges with large choroidal vessels visible below), and elevated plasma levels of ornithine ten to twenty times higher than the normal concentration. Treatment consists mainly of dietary restriction of arginine (ornithine precursor amino acid) such as nuts, seeds, cereals, dairy products, seafood, meat, chicken, watermelon and chocolate and supplementation of vitamin B6 to increase enzyme activity. [2].

Case presentation

This is a 53-year-old patient, with a history of Diabetes Mellitus for 23 years with adequate treatment (fasting glucose of 98 mg/dl, glycosylated hemoglobin 6.5%). He comes to the ophthalmology consultation with a history of decreased visual acuity for several months.

In an initial examination of the fundus, the ophthalmologist observed changes suggestive of retinal dystrophy. Initial examination showed visual acuity in right eye: 20/50, left eye: 20/50. The examination of the adnexa and the anterior segment in a slit lamp showed no alterations. Color vision explored with the Ishihara test was normal, intraocular pressure was 13.7 mmHg in right eye and 12.7 mmHg in left eye. Pupillary reflexes were normal; ocular movement ductions, versions and vergences were present and orthophoria was in primary gaze position. Slit lamp examination of the adnexa and anterior segment showed no alterations; the media shows opacity. In the fundus of the eye, by indirect binocular ophthalmoscopy, normal-sized and slightly pale optic discs were found, discrete vascular thinning, and confluent areas of retinal and choroidal atrophy in the posterior pole that compromised the macula were shown (figure 1).

Complementary examinations are performed which report retinal atrophy areas in the posterior pole that compromise foveolar area in the retina photo. In the optical coherence tomography study, HUVITZ reports disorder in the outer layers of the retina; in addition, plasma ornithine levels were taken which were 650 nmol/ml.

With all these findings, choroidal gyrate atrophy is diagnosed. She was referred for a nutrition consultation and treatment with oral vitamin B6 was indicated.

Discussion

In its differential diagnosis, some diseases must be ruled out, such as atypical retinitis pigmentosa, paving stone degenerations and degenerative or malignant myopia. [3].

In atypical retinitis pigmentosa: lesions in the form of bone spicules predominate and extend from the periphery

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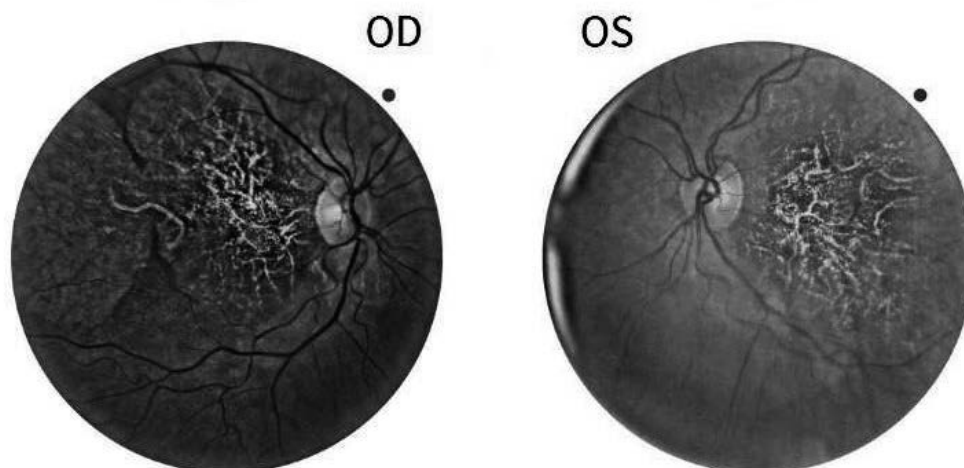


Figure 1. The fundus photo of both eye in patient with gyrate atrophy. The normal-sized and slightly pale optic discs were found, discrete vascular thinning, and confluent areas of retinal and choroidal atrophy in the posterior pole that compromised the macula were shown.

towards the inside as the disease progresses. In paving stone degeneration: lesions predominate in the periphery of the lower quadrants. In cases with extreme degenerative myopia: in cases of severe malignant myopia there are lesions in the posterior pole. These are multiple independent, full-thickness atrophic lesions. But they do not follow the pattern of gyrate atrophy [4, 5].

Treatment consists of 3 pillars: increasing pyridoxine levels, decreasing protein intake, and preventing eye complications.

The importance of a pyridoxine (vitamin B6) supplementation that can vary from 15 to 750 mg/day is reported. Some studies report long-term treatment with Vitamin B6 with a dose of 300 mg/day for 6 months. It should be kept in mind that there are some medications that can lead to a decrease in pyridoxine such as some anticonvulsants. Secondly is a restrictive arginine diet (low protein diet 25g / day) may decrease ornithine to normal levels but only twenty percent of patients tolerate this diet [7, 8].

Patients who respond favorably to vitamin B6 therapy, when compared to those who do not respond, will have a mild to moderate course of the disease with better visual function and chorioretinal lesions will be less extensive. Although one study reports that less than 5 % of the cases diagnosed with GA, with confirmed biochemical tests, had a good response in diet therapy with vitamin B6.1 supplementation.

In conclusion, the following details should be taken into account: a diagnosis of GA at an earlier age slows down the progression of the disease and the appearance of visual complications. Because of the infrequency of the disease, it can go unnoticed for years. Its biochemical confirmation must be carried out. Examination of the rest of the family is essential, as this disease has an autosomal recessive hereditary pattern.

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Disclosures

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