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Commentary

Fundus Autofluorescence Assessment in Macular Disorders

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DESCRIPTION

Stargardt disease is the most common form of Macular dystrophy. The majority of the time, it is inherited as an autosomal recessive trait. This disease's vision loss usually affects young people. Stargardt disease affects about one person in every 10,000, and it usually affects adolescents and young people under the age of 20. Stargardt disease and fundus flavimaculatus are two clinical manifestations of the same disease. A histological accumulation of lipofuscin-type material occurs in the cells of the retinal pigment epithelium due to ABCA4 gene mutation. This mutation is inherited when both parents have a gene malformation. Stargardt disease causes blurry, lacklustre vision, which makes it difficult to recognise faces and shapes as well as read from close up and from a distance, and it eventually leads to colours of similar shades being confused with each other. It also makes it difficult to adjust to the shade. Although it does not result in total blindness, those affected may lose visual acuity to the point where they are legally blind. Patients with this pathology are classified into four groups.

The first stage of the disease is distinguished by the beaten bronze colour of the ocular fundus and choroidal silence. Except for the typical "silent" (dark) choroid that shows up on fluorescein angiography, the back of the eye is almost normal. In the second group, atrophic maculopathy with or without yellow flecks, the loss of RPE may be so minor at first that it only becomes apparent during an angiogram in some patients. Furthermore, the bronze fundus and silent choroid may not be visible in the first decade of life. However, later yellowish lesions appear, indicating lipofuscin storage. The degree and pattern of macular atrophy vary and do not always correlate with the degree of vision loss. A doughnut pattern is a very common pattern that appears, which in some cases allows visual acuity to be preserved until the age of 40. These patients may develop sub-retinal neo vascular membranes that cause disc form lesions on the macula on occasion. The colour vision test usually reveals a slight shift on the red-green axis. Many patients have long-term dark adaptation.

Patients in the third group have atrophic maculopathy with late signs and symptoms of Retinitis pigmentosa. These are similar to the second group, but at a later age, symptoms and signs of Retinitis pigmentosa appear, such as night blindness and abnormalities of the photopic ERG (in bright conditions, resulting in cone dysfunctions) and the scotopic ERG (in dark conditions and entailing rod dysfunctions). Currently, it is thought that these patients have cone and rod dystrophy caused by severe ABCA4 mutations. The fourth group of patients has yellowish flecks that are not associated with macular atrophy. It is the clinical syndrome known as fundus flavimaculatus. These patients may have central and paracentral yellowish lesions with no evidence of RPE atrophy between these lesions on angiography or fundoscopy. Usually, there is choroidal Visual acuity may be normal if the center of the fovea is not affected by one of these lesions, although many patients have a large fleck in the foveola and reduced visual acuity. In the absence of information on the rest of the family and in eyes without very clear evidence of choroidal .It may be difficult or impossible to distinguish them from a dystrophy in a pattern resembling "fundus flavimaculatus."

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