What You Should Know About Congenital Achromatopsia

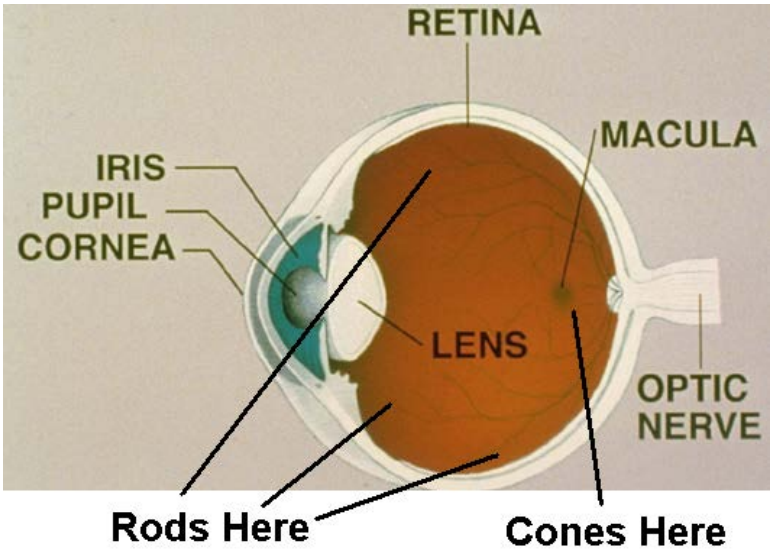
David J. Browning, MD, Ph.D.

Congenital achromatopsia is a condition in which one of several genetic mutations causes the cones of the retina to develop improperly. Patients with congenital achromatopsia have problems with color vision, have decreased visual acuity, are bothered by bright lights, and sometimes have rhythmic eye motions called nystagmus.

The Retina

The retina is the lining of the back of the eye upon which the light is focused. It has two types of cells called cones and rods. The cones are responsible for color vision and fine central vision (the perception of details, reading, etc.) The rods are responsible for peripheral vision. Figure 1 shows that most of the rods are in the peripheral retina. Most of the cones are in the central retina. Cones work under conditions of bright light. The rods work in dim light. In congenital achromatopsia, the cones are dysfunctional or missing, accounting for the prominent problems with day, color, and central vision. The problem is present from birth, tends to be static, and is rare, occurring in approximately 1 in 33,000 births.

Fig. 1



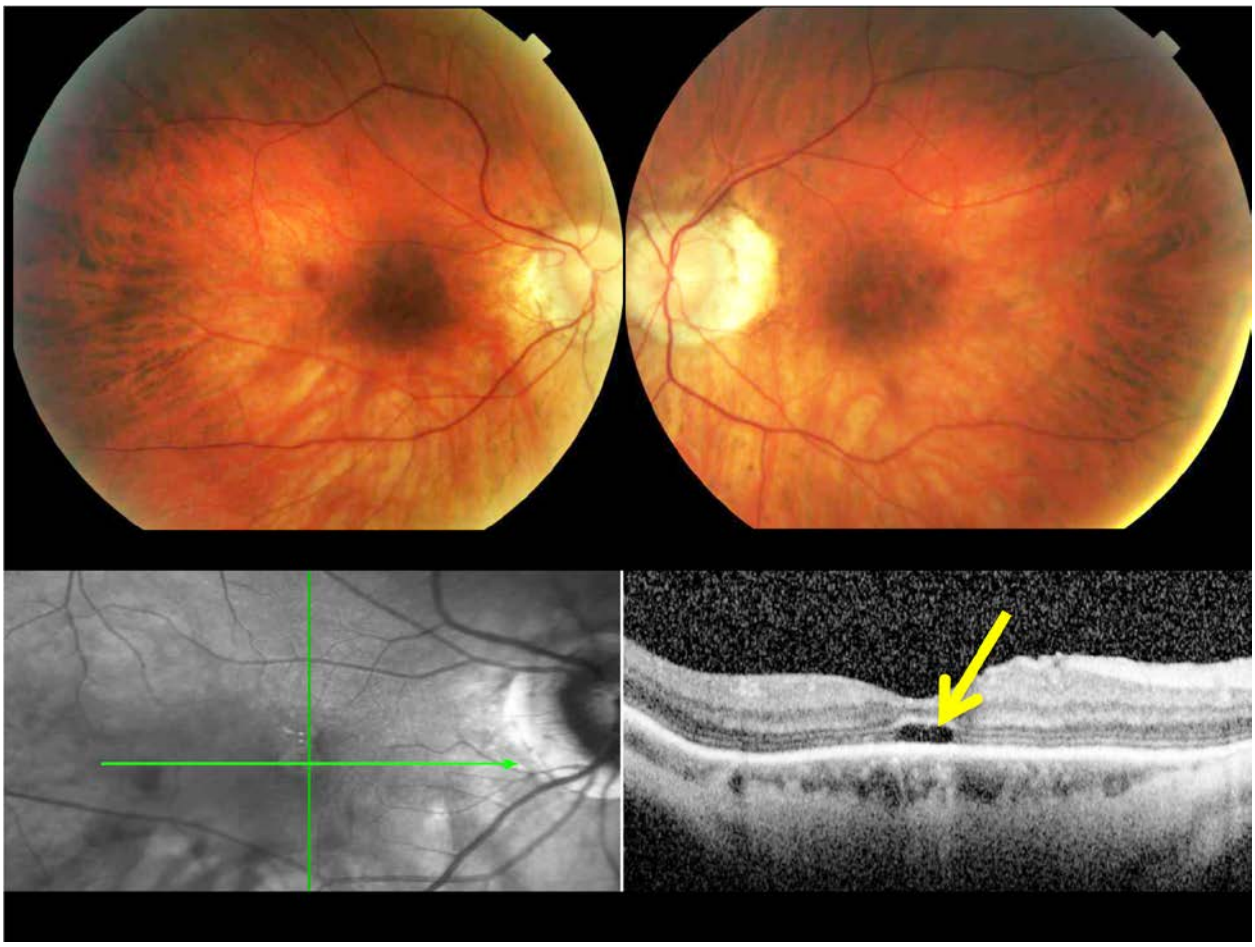
Genetics of Congenital Achromatopsia

Congenital achromatopsia is a disease caused by a gene defect. That is, one is born with the disease and cannot acquire it from the environment. Most cases of congenital achromatopsia have a recessive form of inheritance. This means that a person must have two defective genes to have the disease. Each parent must contribute a defective gene to the affected patient, but in almost all cases neither parent has the disease, because each of them has only one copy of the defective gene. Since it is rare for two carriers to mate, the physician often seeks a history of consanguinity between parents of the affected person. That is, are the parents perhaps distantly related as a way of accounting for the fact that they both have the same defective gene? If an affected patient marries an unrelated person, the chances of any child having the disease are nearly zero. On the other hand, all the children would carry the recessive mutated gene. A number of genetic mutations have been found in various pedigrees of congenital achromatopsia. For example, defects in the CNGA3, CNGB3, and GNAT2 genes have been described, and probably more will be discovered.

Diagnosing Congenital Achromatopsia

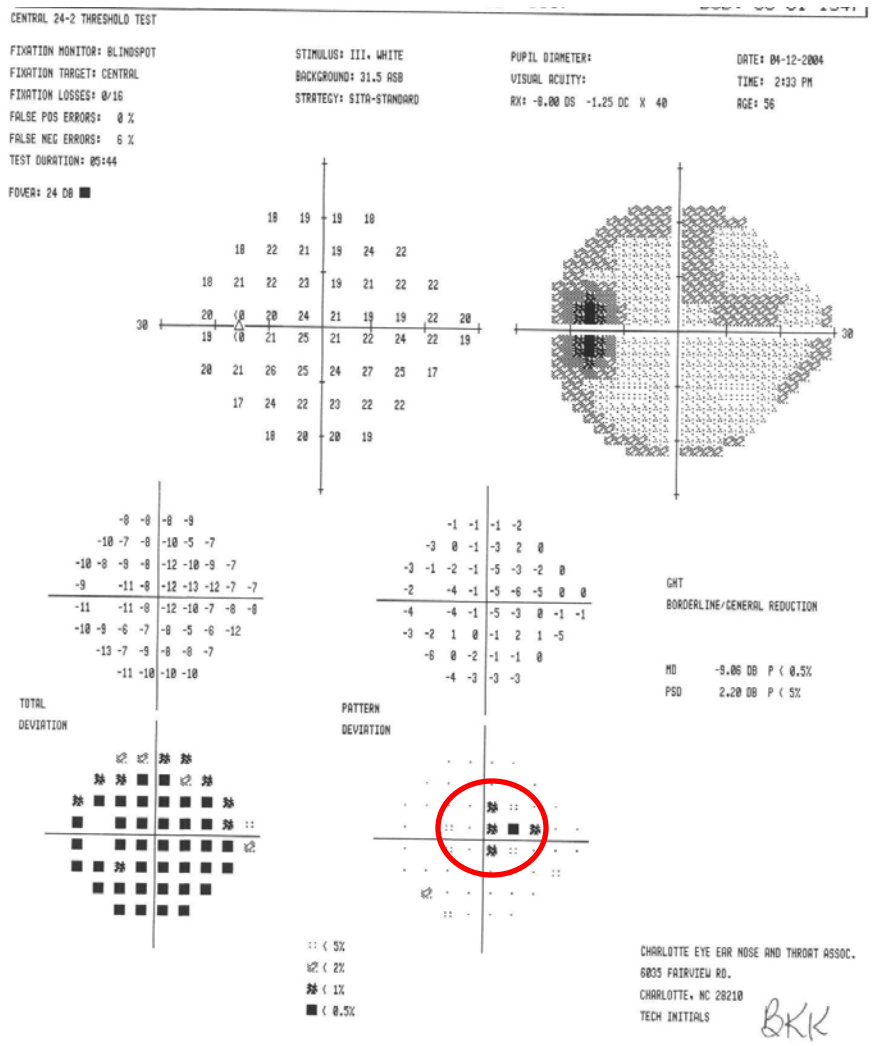
A detailed history of symptoms and a detailed family history are taken, followed by a complete eye examination, which requires dilation of the pupils and use of the slit lamp, headlight, and various lenses. Ancillary testing including electroretinography and spectral domain optical coherence tomography are helpful in making the diagnosis. Genetic testing may be done based on a vial of blood. The findings in an affected patient are shown in figs. 2-4.

Fig. 2



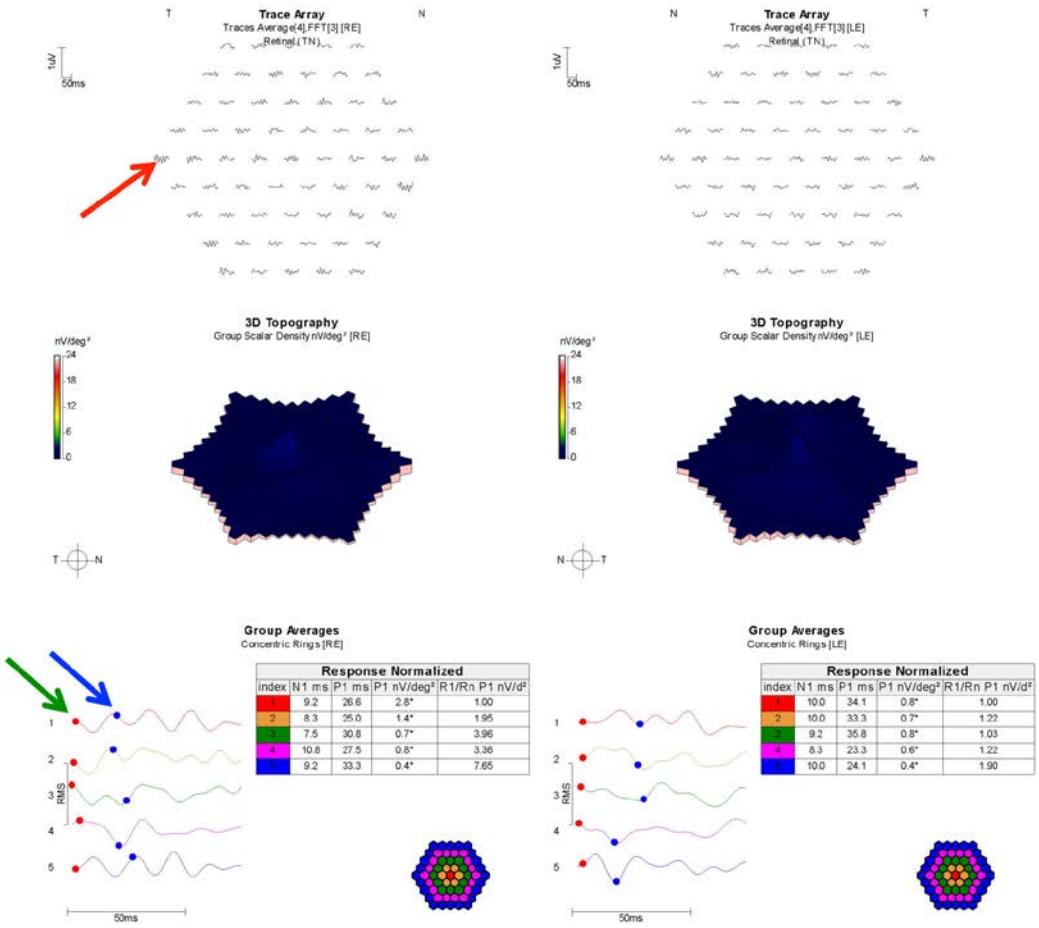
Legend: Fundus photographs (top panel) of a 65 year old man with congenital achromatopsia and visual acuity of 20/80 right eye, 20/100 left eye. The maculas have mild pigment mottling. The bottom panel shows a spectral domain OCT image of the right macula. The yellow arrow shows a cavity where normally cones are found.

Fig. 3



Legend: Visual field of the left eye of the patient shown in fig. 2. There is a central blind spot (red ringed area).

Fig. 4



Legend: Multifocal electroretinogram of the eyes of the patient shown in fig. 2. The waveforms are flat in both eyes. The red arrow shows some 60 cycle noise. The blue and green arrows show that the automatic cursor placement is erroneous because the waveforms are so low in amplitude.

Treatment

At the present time there is no treatment for congenital achromatopsia although active research is ongoing. Patients can contact the National Society to Prevent Blindness (Schaumburg, IL) for educational materials. Low vision aids are useful including magnifiers, closed circuit TVs, and talking books. Most cities have a clinic specializing in low vision evaluations for patients referred by area ophthalmologists and optometrists. Fortunately, the disease is generally nonprogressive; it never causes loss of peripheral vision.

After reading this brochure, if you have a desire to read in more depth about congenital achromatopsia, an excellent resource is the Pubmed page on the National Library of Medicine website at the following link: **http://www.ncbi.nlm.nih.gov/entrez/query.fcgi.**

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