

What You Should Know About Coats Disease

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Coats disease is a congenital condition in which abnormal blood vessels in the retina leak and fail to properly nourish the tissue. The retina is the neural lining of the back of the eye that converts light to a nerve signal. The condition is usually nonhereditary (meaning not passed from generation to generation) however recent investigations have revealed several cases linked to a genetic defect within the NPD gene located on the X Chromosome (Xp11.4). This gene encodes a protein called norrin, which is critical in normal blood vessel development. The linkage to the X chromosome explains why males are more commonly affected by Coats disease (76%). No other organ systems are involved and usually only one eye is diseased (95%), all for reasons unknown. Most patients are discovered before the age of twenty, although sometimes a patient is initially diagnosed later in life, perhaps because of milder disease and late decompensation. To use as a basis for comparison, figure 1 shows the appearance of a normal retina. Figure 2 shows the appearance of the retina in Coats disease. Dilated, sausage-like vessels are present. Fluorescein angiography, a series of pictures taken after the injection of a food coloring into the veins, will often show areas of the retina that lack blood supply. At the borders of such areas, fronds of abnormal elevated vessels may grow into the vitreous gel, which fills the eye. These may lead to scar tissue or bleeding with loss of vision.

Figure 1. Normal Retina

Figure 2. Retina with Coats disease



Clinical Presentation

Since Coats disease frequently involves young people, it is often not recognized by the patient. Frequently, a yellow reflex from the pupil (called leukocoria), or a turning out or in of the eye (called strabismus) will alert a parent or teacher to an ocular problem leading to a visit to the ophthalmologist. By definition, patients with Coats disease do not have any other health problems, but occasionally a retinal picture suggestive of Coats disease will appear as one aspect of a multisystem disorder. For example,

patients with Fascioscapulothoracic Muscular Dystrophy, Pericentric Inversion of Chromosome 3, and Alport Syndrome have been described with a Coats disease-like picture. In these systemic conditions, Coats disease may be more often bilateral. A case of bilateral Coats disease should prompt a search for an associated systemic condition.

Sometimes Coats disease resembles more serious conditions, such as retinoblastoma, which is potentially life threatening. It is critical that an accurate diagnosis be made, as treatment of these two conditions is very different. Thus, ophthalmologists experienced in managing Coats disease should be involved in decision making for all patients.

Treatment of Coats Disease

Treatments for abnormal vessels include laser photocoagulation and cryotherapy (freezing). In both techniques, the abnormal blood vessels are destroyed. Usually laser treatment is chosen for milder cases and cryotherapy for more advanced, severe cases. Often, multiple sessions of treatment are necessary. The usual pattern is to have a treatment, then look at the response in 2-4 months to determine if further treatment is necessary. In extreme cases, vitrectomy surgery may be necessary. This involves the removal of the vitreous fluid and drainage of large fluid collections under the retina. Rarely, a blind, painful eye may need to be removed (enucleation).

Even after successful therapy, patients with Coats disease require lifelong follow-up and sequential examinations because of the tendency for leakage to recur. Use of safety glasses is recommended whenever patients engage in active sports, shop work, or yard work.

Prognosis

Because patients are frequently diagnosed with advanced disease, it is not unusual for retinal scarring to persist, even if retinal swelling and subretinal fluid resolve. In the largest subclinical series reported to date, only 14% of eyes ended with driving vision or better, greater than or equal to 20/50.

Final Comments

Coats disease is a serious eye disease frequently diagnosed at an advanced state. Often, repeated treatments are needed to stabilize affected eyes, and lifelong, serial monitoring is required. Careful distinction of Coats disease from retinoblastoma is important.

After reading this brochure, if you have any questions, please call our office, at 704-295-3182. If you are interested in learning more on your own, we have developed a website dedicated to educating people about retinal disease. Our site is called *The Retina Exchange* and it can be accessed at: www.theretinaexchange.com. This site includes numerous other pamphlets, all written by Drs. Browning and Sanders, which

discuss retinal disease and treatment. There is also a forum available on the site, where patients can read about the experiences other patients have had and share their own experiences if they choose to do so. An additional resource we recommend is the website for the National Library of Medicine, on which there is a diverse collection of medical publications. We have included a link to this site on our website, but it can also be directly accessed via www.pubmed.com.