Incidentally discovered optic nerve head drusen

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A 34-year-old white woman presented for holocephalic headaches following a blow to the head. She did not mention any vision disturbance. Computed tomography was performed and displayed bilateral calcific deposits in the disk optic areas consistent with optic nerve head drusen.

Comment

Optic nerve head drusen (ONHD) is an uncommon condition, resulting from intra- and extracellular mucoproteinic deposits in the optic nerve. These deposits, called "drusen bodies", gradually enlarge and tend to calcify. Other threatening ocular conditions seem to be associated including vascular occlusions, choroidal neovascular membrane (CNVM) and retinal hemorrhages.

The prevalence of the disease in the adult population is about 0.34% and can reach 3.4% in individuals with a family history of ONHD suggesting an autosomal dominant inheritance pattern with incomplete penetrance. Drusen bodies are usually bilateral (70-80%) and more commonly affect Caucasians. There is no gender predilection.

Slowly progressive peripheral visual field loss is the main symptom and occurs in 75% of patients. In contrast, central visual loss seems to be rarer and needs to rule out other causes such as tumor.

Diagnosis of superficial drusen is generally made with dilated fundoscopic examination. ONHD typically shows yellow bumpy deposits over the optic disc and often gives the appearance of an edematous optic nerve with abnormal vascular patterns. B-scan ultrasonography and computed tomography (CT) are useful modalities to detect deeply buried drusen. The main differential diagnostics are true papilledema, anterior ischemic optic neuropathy, calcified granuloma, astrocytoma and optic nerve glioma.

On CT imaging, drusen appear as rounded calcifications in the superficial layers of the optic disc (Fig. A, B). CT presents several disadvantages: exposure to ionizing radiation, high cost and the risk of false negatives. This is due to the tiny diameter of drusen ranging from 0.05 mm to 0.75 mm. Thus, thin-slice CT images are required. B-scan is clearly superior to highlight ONHD but CT remains interesting if clinical presentation leads to a suspicion of intracranial lesion.

Currently, there is no treatment for progressive vision loss due to ONHD. Some clinicians prescribe ocular hypotensive agents in order to prevent further optic nerve damages. With regard to CNVM, different treatments have been reported: laser photocoagulation, photodynamic therapy and surgical removal.

Reference


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