

# NANOS Patient Brochure Drusen

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### **Optic Disc Drusen**

Optic disc drusen are abnormal deposits of protein-like material in the optic disc – the front part of the optic nerve. We do not know the exact cause of optic disc drusen but they are thought to come from abnormal flow of material in optic nerve cells.



Normal Optic Disc



Lumpy Appearance of Optic Disc Drusen



Enlarged view of Optic Disc With Drusen

Optic disc drusen occur in about 1% of the population and are found more frequently in Caucasians. In three-quarters of cases they appear in both eyes. Optic disc drusen may be inherited or may occur without any family history. When drusen are inherited, it is usually as an autosomal dominant trait, which means that it is passed on by one parent, in contrast to some hereditary conditions in which both parents must carry the trait. This means that your children would be at some risk for having the same condition.

Although the tendency to form optic disc drusen is present at birth, the deposits don't actually become visible for several years. The drusen tend to develop slowly over time as the abnormal material collects in the optic nerve head and calcifies. The average age when optic disc drusen first appear is about 12. Often, the optic disc has an unusual appearance, with multiple branches of the major blood vessels as they emerge on the disc.



Multiple Branches of Blood Vessels

Over time, optic disc drusen tend to calcify and thus become more prominent. Drusen are rarely associated with any systemic disease or eye disease.

#### Symptoms

Optic disc drusen often come to medical attention during a routine eye examination. Patients usually have no symptoms and do not notice any problem with their vision. Occasionally, patients may have flickering or graying out of vision that lasts a few seconds or they may notice missing areas of vision. The elevation of the optic disc caused by drusen may be mistaken for papilledema, which is swelling of the optic nerve from increased pressure within the fluid around the brain. Thus, drusen are sometimes referred to as "pseudopapilledema". Because true papilledema is considered a medical emergency, patients with disc drusen are sometimes referred for further testing, to a neurologist, neuro-surgeon, or even the emergency room, on an urgent basis.

#### Diagnosis

Optic disc drusen may be buried within the substance of the optic nerve head or located superficially on its surface. When superficial, they appear as glistening yellow bodies and can be seen more readily by ophthalmoscopic examination. When buried under the surface, drusen are hidden from direct view and diagnosis is more challenging. As drusen become calcified, they can be detected with ultrasound, optical coherence tomography (OCT) and occasionally by CT scanning.



Ultrasound showing large druse

#### Prognosis

Most patients with optic d is c drusen retain normal central vision. However, over time up to 70% of patients may lose some peripheral vision. This kind of visual loss is picked up by visual field testing, also termed <u>perimetry</u>. The amount of peripheral visual field loss varies from none to severe constriction. Patients with optic disc drusen may also be at an increased risk for developing non-arteritic anterior ischemic optic neuropathy (NAION), branch retinal vein occlusion (BRVO), and central retinal vein occlusion (CRVO).

#### Management and Treatment

There is no proven or standard treatment for optic disc drusen. Nevertheless, careful monitoring of the visual field is recommended to detect progression of visual field loss. Rarely, a small area of new blood vessels called a choroidal neovascular membrane [CNV] can develop adjacent to the optic disc. A CNV has a tendency to bleed and cause sudden visual loss. Early detection of the presence of CNV is extremely important because prompt treatment can often prevent serious complications from bleeding.

#### Frequently Asked Questions

#### Why did I develop optic disc drusen?

Optic disc drusen are caused by an abnormal deposition of a protein-like material in the optic nerve. The cause of this material is unknown. In some individuals, the deposition of this material can be inherited, while in others it occurs without a family history.

#### How does my doctor diagnose this condition?

Your doctor can diagnosis this condition either by an ophthalmoscopic examination or with the aid of ultrasound, optical coherence tomography (OCT) or computer tomography (CT) scanning.

#### Will the drusen get worse?

The number and size of the optic disc drusen tend to increase over time.

#### Can this condition affect any of my family members?

Yes, optic disc drusen can occur as an inherited family trait and may affect close relatives. Patients diagnosed with optic disc drusen should consider discussing their diagnosis with their family members so that they undergo screening evaluation. Optic disc drusen usually do not show up in Infants and children younger than 4 years.

## Should I let other physicians who may be caring for me or other family members know about my condition?

Yes, it would be helpful for other physicians who are involved with your care or the care of a family member to know that you have diagnosed with optic disc drusen. You should inform them that you do not have papilledema. Is there anything I can do to prevent the drusen from getting worse?

No, there is no proven way to prevent the optic disc drusen from increasing in size.

Is there any treatment for this condition?

No, there is no proven treatment for optic disc drusen at this time.

If there is no treatment for optic disc drusen, why should I have regular ophthalmic examinations?

Occasionally, a patient with optic disc drusen may develop new abnormal blood vessels (choroidal neovascularization) adjacent to the optic nerve. These vessels are fragile and prone to bleeding which can damage the adjacent retina. This kind of damage can often be prevented with laser treatment. Periodic examination is also recommended to monitor for the development of other vision threatening conditions such as glaucoma