Primary open-angle glaucoma: the importance of family history and role of intraocular pressure

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Clinical records

Patient 1

In March 2005, a 58-year-old man was referred by his general practitioner for evaluation of a central scotoma with macular sparing in his right eye of 6 weeks' duration, noticed during reading as a sort of shadow following his point of fixation. He had had no previous ophthalmic complaints and had worn spectacles to correct moderate myopia since the age of 12 years. His health was excellent; he was on no medication and had never had a surgical procedure. Both parents had lost vision from glaucoma. For this reason, his intraocular pressure (IOP) was monitored by his brother, an optician. Over 4 years, repeated IOP measurements of under 22 mmHg had been obtained by non-contact tonometry in both eyes.

On examination, visual acuity was 6/6 in both eyes. IOPs of 38 mmHg in the right eye and 30 mmHg in the left eye were obtained by applanation tonometry. A non-contact method yielded pressures of 28 mmHg in the right eye and 22 mmHg in the left eye. Further examination showed open anterior chamber angles and a myopic optic nerve head which was pathologically cupped on the right side and "suspicious looking", with elongated vertical cupping on the left (Figure A).

Later, automated visual field testing showed a large central defect in the right eye, almost reaching the point of fixation (Figure B). The left eye was normal. Therapy with eye drops (timolol/dorzolamide fixed combination, twice daily) was started, which stabilised the pressure at around 10 mmHg in both eyes.

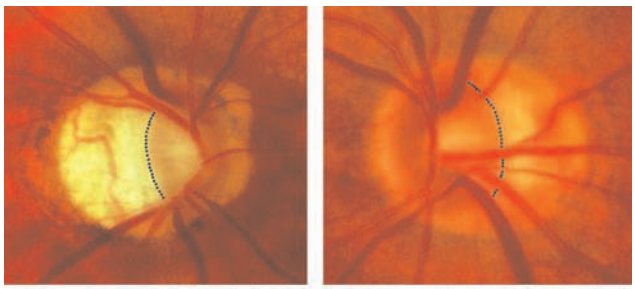
The patient's brother, who had never consulted an ophthalmologist, was advised to do so. He showed no signs of glaucoma.

Patient 2

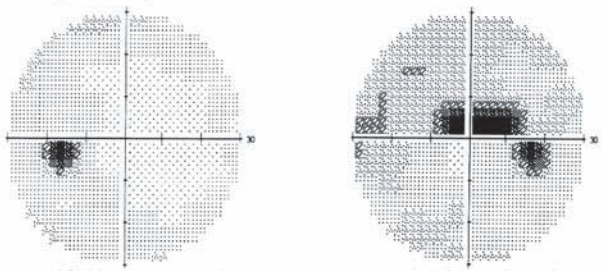
A 69-year-old man presented in June 2005 with slowly deteriorating vision over several years. For the previous 2 months, he complained of bumping into objects in his path. He had never experienced eye pain and had been healthy all his life. His mother, whom he had accompanied to the GP and ophthalmologist for many years, had eventually become blind from glaucoma when very elderly. His sister went blind from glaucoma at the age of 65 years. Twenty-five years earlier, the patient had been seen by an ophthalmologist who saw no signs of glaucoma during a routine examination. The patient claimed that he had never been told to have regular eye checks for glaucoma, and had not done so.

On examination, visual acuity was light perception in the right eye and hand movements at 2 m in the left eye. IOPs by applanation tonometry were 53 mmHg for the right eye and 49 mmHg for the left eye. Gonioscopy showed open anterior chamber angles. Both optic nerve heads were completely cupped (Figure C). Visual field examination showed no vision in the right eye and a temporal island of vision in the left eye (Figure D).

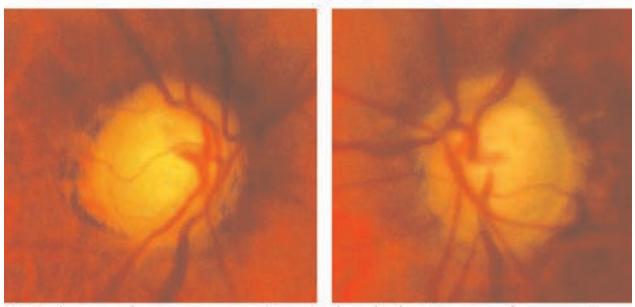
In an attempt to preserve his remaining vision, medical therapy (timolol/dorzolamide fixed combination, twice daily and latanoprost, daily) was instituted, which stabilised pressures at below 18 mmHg in both eyes. He has noticed no further decline in vision since presentation.



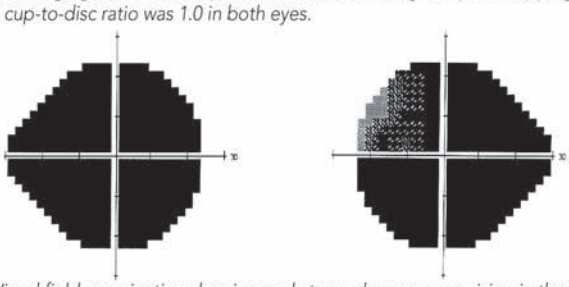
*A: Myopic optic nerve heads showing typical peripapillary atrophy (temporal of brackets). The right eye (**left**) shows advanced cupping, especially at the inferior border. The optic nerve head in the left eye (**right**) looks suspect (notching) at the superior border.*



*B: Visual field examination showing a serious central defect in the right eye (**left**), no defects are shown in the left eye (**right**).*



C: End-stage glaucomatous optic nerve heads showing complete cupping. The cup-to-disc ratio was 1.0 in both eyes.



*D. Visual field examination showing end-stage glaucoma: no vision in the right eye (**left**) and a temporal rest of vision in the left eye (**right**).* ◆

The two cases we present here are good examples of how the diagnosis of glaucoma is often missed because of lack of history-taking and/or insufficient clinical examination. Glaucoma is the second leading cause of blindness worldwide;[1] nevertheless, knowledge of this disease is poor among the general population as well as among health professionals.[2] Primary open-angle glaucoma (POAG) is a chronic and progressive optic neuropathy that causes visual field loss, eventually leading to complete

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blindness. It can be present for a long time before patients have symptoms. About half of those affected with glaucoma are not aware they have the disease.[3]

While the most important risk factor for POAG is elevated intraocular pressure (IOP), pressure is only one component of a constellation of findings that define POAG. Glaucoma is a specific optic neuropathy, characterised by optic nerve head cupping and usually associated with (arcuate) visual field loss. Glaucoma may be associated with elevated IOP, but can be diagnosed irrespective of the IOP, so IOP is no longer considered part of the diagnosis.

Established risk factors for POAG (besides elevated IOP) are higher age, African ethnicity, a positive family history, thinner corneal thickness, and myopia.[1] Diagnostic clues include gonioscopy (visualisation of the drainage angle), characteristic changes in the optic nerve head (cupping — elongation of the optic cup in a vertical direction, as result of, typically inferior, notching of the neuroretinal rim that will be the first sign of POAG), and characteristic (mostly midperipheral) defects on visual field testing. To date, the only effective evidence-based treatment for glaucoma is decreasing IOP;[4] any local or systemic risk factors should also be addressed.

It is critical to note that symptoms occur late in POAG. If a diagnosis is based only on symptoms, advanced irreversible vision loss is likely. Patients with POAG need to be identified early, which requires a high level of suspicion, adequate screening and prompt referral.

In many countries, primary eye care is provided by general practitioners and eye specialists. In others, this role is fulfilled by optometrists. Unfortunately, many health care workers, including (in our experience) even some ophthalmologists, only look at IOP when considering glaucoma as a diagnosis, as in our Patient 1. Over a third of patients with glaucoma have an IOP within the average range (10–21 mmHg).[5] They have "normal tension glaucoma" in which, despite an IOP that is not elevated beyond the arbitrary upper limit of 21 mmHg, the optic nerve can show pathological cupping, and the visual field examination can show characteristic defects. These patients will easily be missed if diagnosis is based on IOP. Moreover, some health care workers tend to use non-contact tonometry to measure IOP. Such tonometry tends to underestimate the true pressure, which is more accurately measured with a Goldmann applanation tonometer (Haag–Streit International, Koeniz, Switzerland).[6] We emphasise that examination of the optic disc is much more important for a correct diagnosis of POAG than IOP.

However, measuring IOP remains important, as our Patient 2 would have been diagnosed by screening IOP alone. We also emphasise that pain is seldom a symptom of POAG. Pain is a feature of acute angle closure glaucoma, in which the IOP reaches very high levels in a very short time because of acute blockage of the drainage angle; this is not a feature of POAG.

Family history played an important role in both our patients and should have been investigated during history-taking. First-degree relatives of patients with POAG have a risk about 10 times greater than for people with no family history of glaucoma.[7] However, relatives are often unaware of their risk, sometimes even decades after treatment is initiated in their family.[8] Patients with primary glaucoma should be advised to alert relatives to the need for adequate glaucoma screening and follow-up.

We believe awareness of POAG among the general population (as well as among health care professionals) is poor at best. To increase awareness in the general population, primary care health professionals in particular need a better understanding of this disease. We believe that asking about a family history of glaucoma as a part of an ophthalmic history, or as part of a general medical history, should be routine in all new patients. First-degree relatives of patients with POAG should be advised to be screened by an ophthalmologist or optometrist. Depending on ophthalmic findings, age and other risk factors, first-degree relatives should have a full ocular evaluation on a regular basis (eg, every 2 or 3 years, or more frequently if findings are equivocal). This simple step, and a timely referral can prevent much disability and associated cost for individual patients and the community.

Lessons from practice

- All primary care doctors should ask their patients about a family history of primary open-angle glaucoma (POAG), which is an important risk factor for this disease.
- Patients with POAG should be told to alert their first-degree relatives to the need for an adequate glaucoma screening.
- "No visual symptoms" does not equal "no POAG", as visual field defects occur mainly at advanced stages of the disease.
- Pain is seldom a symptom in POAG (it is associated with acute angle closure glaucoma).
- About a third of patients with POAG have so-called "normal tension glaucoma"; this will not be detected by measuring intraocular pressure alone.
- Examining the optic disc is much more important for a correct diagnosis of POAG than measuring intraocular pressure. ◆

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Competing interests

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