Childhood Glaucomas and Pseudoglucomas

Instructor:

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Section:

Glaucoma

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COURSE DESCRIPTION:

This course will cover causative factors in congenital and early-onset forms of glaucoma, in order to prevent vision loss and legal blindness later in life in this young population.

LEARNING OBJECTIVES:

1. To discuss the predisposing anterior segment risk factors for juvenile glaucoma
2. To discuss surgical and topical pharmaceutical treatments for these conditions
3. To differentiate megalopapilla, optic pits and colobomae from early-onset glaucoma
4. To differentiate between optic nerve colobomae and myopic staphylomae

(Course begins on page 2)
Welcome to Childhood Glaucomas and Pseudoglaucomas. I am Dr. James Kundart, associate professor at Pacific University College of Optometry and for the next 50 minutes or so, we will be discussing the secondary glaucomas that are often inborn and what looks like glaucoma in children. This is my specialty – pediatric optometry, and I open with a picture of the morning glory flower because the last thing we’ll cover today is Morning Glory Disc Anomaly, which certainly looks anomalous, and is so, but can resemble an optic nerve anomaly like the optic neuropathies caused by glaucomas in children.

For those taking the continuing education exam, here are the learning objectives: (also stated on the first page of this course)

1. To discuss the predisposing anterior segment risk factors for juvenile glaucoma
2. To discuss surgical and topical pharmaceutical treatments for these conditions
3. To differentiate megalopapilla, optic pits and colobomae from early-onset glaucoma
4. To differentiate between optic nerve colobomae and myopic staphylomae

For those taking the continuing education course for credit, you will need to answer 10 multiple-choice questions at the end, and these are 4 learning objectives that are more at the end of the course for you to study. We will be talking about things that predispose children and juvenile patients to anterior segment anomalies that lead to glaucoma, we will also be talking about how these are treated – mostly surgically but sometimes pharmaceutically. We will be differentially diagnosing large optic discs, optic pits and colobomae, and also talking about staphylomae from myopia and other causes.

I took interest in this topic because my family is Slovak Roma, otherwise known as Gypsy, and while I was taught in optometry school that pediatric and childhood glaucomas are rare, happening in only 1 of 20,000 patients, in my ethnic group, it happens that they are 8 times more common (1:2500). Similar high numbers can be found in other parts of Eastern Europe, where groups of the population are predisposed to childhood glaucoma. The question came to my mind of, “Why would this be the case?” The answer is often that it’s a genetic predisposition in the anterior segment that leads to problems with the anterior chamber angle. At first I thought it would be narrow angles running in the family – my grandmother had that, but it seems to be instead open angles that are somehow otherwise anomalous.

The first step in detecting pediatric glaucoma is to convince yourself that it is not exceedingly rare, meaning that you’ll never see it.

If we go back to embryogenesis, there is a neural crest theory called Neurocrestopathy, in which the secondary glaucomas...
originate in tissues that are derived from neural crest cells. Shown in Figure 1, when the neural tube is first forming very early on in the first trimester of gestation, this is where the problems arise. We know that there are midline facial feature problems in some things we’ll see today, such as Rieger Syndrome or Axenfeld-Rieger Syndrome. There is some reason to believe that this theory would be the case.

I should mention for those that are needing to brush up on their ocular anatomy or their embryology that neural crest cells are transient. They are what’s called ‘multipotent’ migratory cells so they can make a lot of things. They are a lot like stem cells in that way. They only exist in the vertebrate animals and people, and they give rise to melanocytes, cranial/facial cartilage, bone, smooth muscle, and also peripheral neurons and glia.

Figure 2 is an electron micrograph of an enucleated eye, and we can see some features labeled here if we look carefully. We see a developing eye surrounded by mesenchyme of neural crest origin. L in the very center is the embryonic lens. H is the Hyaloid Artery, and R is the retina which looks more like iris in this particular picture. P is the Retinal Pigment Epithelium (RPE) in the periphery.

The Neuralcrestopathy theory, while logical, does have some flaws in it. There are many other parts of the midline facial features that are unaffected even in secondary glaucomas. There aren’t necessarily cranial-facial disorders, for example, in these conditions. Even in the eye that has glaucoma, the choroid and sclera, as we can see in Figure 2, are often unaffected. One thing we can say for certain is that neural crest cells move, they migrate in embryogenesis, and they are also affected by their neighbors. So there is some kind of interaction going on here.

The other thing we need to remind ourselves of in embryogenesis, before we move on to patient cases and such, is the fact that there is a tunica vasculosa lentis that surrounds the embryonic lens. (Fig 3) This is a network of blood vessels that come from the hyaloid artery that dissolve in utero when all goes well. We know that you can get Bergmeister’s Papillae, Mittendorf Dot, and vitreal floaters from the pieces of the dissolved hyaloid artery. Sometimes this tunica vasculosa lentis does not completely regress, and what you are left with is at least a persistent pupillary membrane, if not further disorders of the anterior segment. Regression should occur normally within the second trimester.
Figure 4 is the type of picture I had in mind at the time of graduation, some 15 years ago, of what a pediatric glaucoma patient would present as if I were to get lucky enough to see one at all. (If you can call that lucky.) The patient would have steamy, edematous megalocornea, as seen in Fig 4 – right. But they would also have photophobia, so they would be squinting (Fig 4 – left). Perhaps they would have epiphora with tears running down the cheeks, and such. This is certainly true, but it not universally true. This textbook case is the worst case scenario for pediatric glaucoma.

Some of the features that can occur, even in some folks with glaucoma are shown in Table 1. These features of the anterior segment abnormalities that can lead to childhood glaucoma. In Figure 5, the arrows are pointing to Haab’s Striae, which are caused by the high IOP in a patient with congenital glaucoma. They are linear, refractile, and protrude slightly into the anterior chamber. They often go in circles or in a horizontal direction.

**Abnormality #1: Haab’s Striae**

Let’s talk about Haab’s Striae a little bit more. In these cases, you can see with Gonioscopy (Fig 6 right), which should be done on all of these patients that allow us to do it, the anterior chamber angle is wide open, has a deep approach, but there is a thickened trabecular meshwork. That is where the iris inserts. This ophthalmologist noted that there appears to be a glistening membrane covering the trabecular
meshwork which is seen during trabeculectomy surgery. In Fig 6 left, we see Haab’s Striae which is really when you get a good corneal exam of these patients in the slit lamp, are the stretch marks that indicate the IOP has been high at one time, if it isn’t now. Sometimes I find it easier on children to get a slit lamp exam than I do to get a good reading of pressure, particularly Goldmann pressure. These striae can lead me to know when I really need to work hard to get an IOP measurement.

Abnormality #2: Axenfeld Anomaly (Syndrome)

![Figure 7: Axenfeld anomaly is anterior displacement of Schwalbe’s line associated with iris strands peripheral to it (Wright, page 47). Picture source: Morrison & Pollack, Fig 17-2.](image)

We are going to split out Axenfeld Anomaly and Rieger Syndrome into their traditional separate definitions here. Nowadays they are recognized as continuum along the same disease spectrum. What we see in Fig 7 left is Axenfeld Anomaly. We see a rather prominent anteriorly displaced Schwalbe’s line, which is where Descemet’s membrane meets the cornea. This patient has Axenfeld Anomaly. The Syndrome is not there unless they also have glaucoma, in my opinion. The iris processes, seen in Gonioscopy (Fig 7 right) extending to Schwalbe’s line, cause this prominent feature. If you have glaucoma as well, then we call this Axenfeld Syndrome.

Abnormality #3: Rieger Anomaly

Axenfeld Syndrome has been combined with the other doctor that discovered Rieger Anomaly, or Rieger Syndrome again when you have an associated glaucoma. Figure 8, from doctors Morrison & Pollack, in the book edited by them, Glaucoma Science and Practice, in Chapter 17 by John Samples, (available at the Pacific Library to current students and faculty) we have an iridescisis that occurs in the Rieger anomaly. The original pupil is up in the upper left at about 10:00 or 11:00, the middle elongated oval. Peripheral to the original pupil at 10:00 or so, we have a little schisis, and then the large schisis creating a larger secondary pupil to the right. This is a sign that this patient is at high risk for glaucoma. In the full-blown Rieger Syndrome, there are microdontia, and hypodontia where teeth are small or don’t form at
Figure 9: Left: Small teeth (microdontia) and unformed teeth (hypodontia) seen with Rieger Syndrome. Right: Iris atrophy

The shape of the anomalous teeth can be seen in Figure 9 (left). Patients may well have an obliquely-shaped cat-like pupil that looks like an iris coloboma, but is, in fact, iris atrophy. (Fig 9 right)

With Axenfeld and Rieger Syndromes, we can also have Posterior Embryotoxon. Notice the spelling – with many O’s. What we have here is something that you are going to see in 10-15% of normal eyes, so 1 out of every 7 to 10 patients is going to have this. I think it’s easier to see on a darker iris. Figure 10 shows us an isolated temporal Posterior Embryotoxon in a normal eye. We have a line parallel to the limbus, running down along the temporal side. This does not increase the risk of glaucoma, but is an anteriorly displaced Schwalbe’s Line, where Descemet’s membrane meets the cornea.

In Axenfeld-Rieger Syndrome, Posterior Embryotoxon may be more prominent. In Fig 11 right we can see engorged vessels and definitely the posterior embryotoxon. It is more faintly see on Fig 11 left, where again there is some anomaly within the anterior chamber angle, even visible from the anterior view of the slit lamp, without a gonio lens. Notice the blood vessels and where they travel to, and how it appears perhaps like neovascularization, but instead it is that the anterior chamber angle is anomalous.

Figure 10: Posterior Embryotoxon

Figure 11: More examples of posterior embryotoxon
Abnormality #4: Peter's Anomaly

The last anomaly usually discussed in this triad besides Axenfeld and Rieger Anomaly is Peter’s Anomaly. This one is harder to miss because there is a central corneal opacity that occurs by an anterior synechiae. Figure 12B has a bubble in front created by the gonio lens. The synechiae creates a loss of endothelial function and loss of endothelial pump function, causing an opaque cornea.

Descemet’s membrane may be absent where the opacity happens. Here we can see in Figure 13 in these enucleated eyes we can see a rather prominent doughnut pattern on the left of leukocoria which suggests Peter’s anomaly. On the right eye, the endothelium has covered the defect and created a new basement membrane. It leaves the corneal opacity, and the edema regresses. 50% of Peter’s Anomaly cases may show bilateral glaucoma, sclerocornea, corectopia or a misshapen pupil. Sometimes we see iris hypoplasia, or anterior polar cataracts. Iris corneal adhesion is present, as we’ve seen. Also we can see kerato-lenticular touch, meaning the lens and the cornea may be touching. Microcornea happens often, as does Aniridia or under-formed iris, hypo-iridia. There can also be a retinal-choroidal coloboma, iris coloboma, and persistent hyperplastic primary vitreous, as well. There are a whole slew of features here, but the reason we don’t see these as often is because they all go to pediatric ophthalmology. If you work in co-management with these doctors, especially in-house, you will see this a lot more often than you ever thought you would.

Figure 14 is an example of sclerocornea. Teratogen exposure in-utero in mice has been shown to cause this. The third week of gestation, if the mother ingests a drug that might lead to birth defects, that teratogen may lead to Peter’s Anomaly in her unborn child, very unfortunately.

A hand-held slit lamp is sometimes used on these pediatric patients. Sometimes they have multiple special needs, such as they may be in a wheelchair and have a
hard time getting behind a slit lamp, regardless of their age. The anterior chamber may have not generated, or maybe have dysgenesis. Figure 14 is a blind eye that is at risk for glaucoma. Even the non-seeing eyes need to be tested for high IOP lest they become painful, and need to be enucleated.

We talked about the iris adhering to the lens, but Figure 15 shows us an example of what happens when the tunica vasculosa lentis doesn’t completely regress in childhood. What you can be left with is a sort of persistent pupillary membrane on steroids, if you will. The steroid analogy is doubly appropriate because in this picture we also have a cataract forming, as well as many, many iris strands probably following the pathway of the tunica vasculosa lentis at one time that has partially regressed, leaving us with these adhesions as a type of posterior synechiae and a cataract, which also leaves the patient at risk for amblyopia as well as juvenile glaucoma. This is an anterior polar cataract, as opposed to the posterior sub-capsular (PSC) caused by steroids.

**Abnormality #5: Aniridia and Nephroblastoma**

We mentioned that Aniridia is also a complication that can occur here. Aniridia is occurring with nephroblastoma in cases of what we used to call Wilms Tumor. This is actually a very treatable tumor – at least 9 out of 10 patients survive at least 5 years. However, it needs to be detected in early childhood, so if you see a patient with even partial iris genesis, and an opaque lens with some abnormal vasculature going over the pupil as we see in Fig 16, this is someone who is at risk for Wilms Tumor. African-Americans are at higher risk for this than Caucasians are, but it does occur in all of the ethnicities, as far as I know. These are children who are preschool age, just the age of my own son, and we know that these two are related because both the kidney and iris formation are encoded on Chromosome 11.

**Abnormality #6: Microcornea**

Going to item #6 in our list of predisposing factors for juvenile glaucoma, Microcornea is seen in Figure 17 on the left eye. This is very frequently associated with fetal alcohol spectrum disorders. It doesn’t have to be the full-blown Fetal Alcohol Syndrome. The spectrum disorders can lead to Microcornea in one or both eyes, and in fact usually is asymmetrical but with both eyes having the problem.

The cornea in a normal adult is about 12mm across, with a little tolerance on either side of that value. I don’t know what you would call a cornea exactly
that is just above 10, but most textbooks define Microcornea as a cornea with a diameter that is below 10mm. 9mm or less in children. You will see it 90% of the time in fetal alcohol spectrum disorders, which is a difficult thing to discuss with parents, especially the birth mother. We need to be aware that when this does occur and we are suspicious of fetal alcohol spectrum disorder, that doesn’t mean that the mother was negligent or was an alcoholic – it has been observed and documented that half the pregnancies in the US are unplanned and half the women of childbearing age admit to drinking, so by simple math, ¼ of the unplanned pregnancies are by women who drink, which means they may have exposed the infant to some amount of alcohol before they knew they were pregnant. The prognosis for surgical reconstruction and repair is guarded, and the patient is at lifelong risk for glaucoma.

Sometimes Microcornea goes with iris coloboma. You can see how small the cornea is in Figure 18, as well as the keyhole-shaped iris coloboma, probably leading to a retinal coloboma, as well. Notice that, unlike Rieger Syndrome, these are usually more or less vertically oriented, like a keyhole, and just a little bit turned to the temporal side.

Table 2

<table>
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<tr>
<th>Microcornea associations</th>
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<td>Anterior segment dysgenesis</td>
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<tr>
<td>Early-onset cataract</td>
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<td>Iris abnormalities</td>
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<td>Childhood glaucoma</td>
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<td>An isolated anomaly</td>
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When you do see an iris coloboma, you want to follow it back and see if there is a retinal coloboma. Sometimes there is trench all the way back from the iris when the neural tube failed to completely close. Other times what you will see is just an isolated off the disk coloboma. Figure 19 (right) shows an off-the-disc retinal and choroidal coloboma. The inferior location is normally where they are. This is called a choroidal skip lesion when it doesn’t quite encompass the whole disc.
You see these in all kinds of combinations. Figure 20 shows a patient with Microcornea of the left eye, at least. Closer examination reveals an iris coloboma in the affected eye, a decentered corectopic pupil, iris pigment on the lens, and a normal right eye. There is a high chance of glaucoma in this left eye. The right eye should be examined as well, not just taken as a control because we may have asymmetrical Microcornea going on here.

Figure 21 is the left retina of the patient shown in Figure 20. In this case, the iris coloboma connects completely to the retinal coloboma, eating up the optic disc. If you can kind of see, just left of center in this picture is where the optic disc should be. The optic cup is very hard to see, perhaps engulfed in the coloboma below. The superior disc is probably intact, leading to inferior visual field being somewhat functional. It’s hard to tell this from glaucoma. How do we monitor for glaucoma other than pressures, when the field and acuities are impaired in this eye? This is a pseudoglaucoma for sure of the back. Thus, anterior segment dysgenesis can lead to posterior segment pseudoglaucoma.

I’ve had the misfortune to see twice in my career so far two cases of extreme microphthalmia or anophthalmia. Figure 22 is a little boy, not one of mine but rather an example from the Wright text, but...
similar to one of mine. This is a 6 month old with colobomatous microphthalmia and orbital cyst anomaly. There is a lower eyelid cyst causing the swelling and edema we see in the lower lid. There is also blepharophimosis and an apparently normal right eye. Figure 22 (right) shows the doctor using a type of retractor here to open the eye lids to expose what generated of the microphthalmic left eye. This is almost an anophthalmia to me – the only remnant that can be seen is the dimple in the center. This is really the optic cup only is what formed in this eye.

We have imaging of this patient, as well. Fig 23 (left) is the CT scan showing the left microphthalmic eye from the bottom, and Fig 23 (right) shows a right optic nerve coloboma. What do you know? The right eye is not normal in this patient, either. The optic nerve coloboma appearing as such, might well appear to be a glaucoma to us, and of course there is risk for both eyes in this particular case. Since there is only one sighted eye, we want to be careful with this. There was a stalk found connecting a cyst in the microphthalmic eye, which was surgically removed.

In the good eye, the right eye, there was a large, isolated, optic nerve coloboma, but otherwise the eye was normal. This image could almost be mistaken for Morning Glory Disc Anomaly, as well.

We have seen now the spectrum now from Posterior Embryotoxon, which can be normal in up to 15% of our patients, to Axenfeld-Rieger Anomaly, Microcornea, to Microphthalmia. All of these cases can cause true risk of secondary glaucoma in childhood. We have to consider what the treatment should be.

**Treating Childhood Glaucoma**

Usually treatment will be surgical, so again, if you are not working with a pediatric ophthalmologist, or perhaps even a pediatric glaucoma specialist in larger cities, you are not going to be seeing this, because surgical treatment is the treatment of choice.

If the IOP is greater than 21 mmHg, as measured by a reliable method (ideally Goldmann, but a handheld Perkins tonometer or the iCare tonometer, which seems to be taking over pediatric optometry) by applanation on two occasions under anesthesia is considered abnormal IOP. In these patients, we want their pressures to be closer to 12, but within the range from 8-9 mmHg to about 16 mmHg. This is a little lower than adults, and often we find that if you take
NCT on children that their trepidation and nervousness causes the NCT IOP to go up due to the tightness of their eyelid when they are awake. Sometimes using proparacaine, if you can trust them to not rub their cornea off, then doing NCT may give you a more reliable pressure.

Trabeculectomy (Fig 24) is often the treatment of choice for these patients. If they happen to have a narrow angle, of course a surgeon can do a peripheral iridotomy.

You may need to use topical medications with the surgery, but there are some you don’t want to use, and one you definitely don’t want to use is Alphagan. Brimonidine is known as a neuroprotective agent, and the reason for that is that it crosses the blood-retinal barrier, which is really part of the blood-brain barrier. The problem with this in seniors is that it causes them to fall asleep in the middle of the day, especially if they are on TID dosing, and you can tell that it’s not just due to their age because if you remove the drop and they don’t take the midday nap anymore. This happened in my grandmother’s case, as well as many patients you may know. In children, this not only knocks them out so that they sleep all the time, but it can cause various other psychiatric disturbances, delirium among them. Thus, this is a contraindicated drug in children.

Often in children, you don’t want to reach for the prostaglandins because you don’t want to risk changing their iris or orbit color or causing them to grow really long lashes, if they’re a boy, especially, and all those sorts of things. Traditionally our second line of defense is the alpha agonists may not be a good one for them, either.

**Pseudoglucomas**

So those are the ‘true glaucomas’ in the first half of our presentation. I’d like to spend the second half of this presentation talking about pseudoglucomas and see where that gets us.

Table 3 shows a list of some pseudoglucomas that can mimic and can occur in absence of the previously discussed anterior segment abnormalities. Notice really everything we’ve seen so far in the true childhood glucomas are secondary to anterior chamber problems. The pseudoglucomas are posterior chamber problems where you look at the optic nerve, which is often hard to get a look at in a young child. When you finally do manage to get a look, you have a megalopapilla, an optic pit, an optic disc coloboma, all of which mimic optic neuropathy due to glucoma. The optic nerve can also be hypoplastic or malinserted and of course we have the Morning Glory Disc at the end to talk about.

These are all failures of the fetal fissure to close fully, and the patient may also have visual field defects, but the difference is that these are not progressive conditions.
Megalopapilla

Figure 26 shows Megalopapilla. Basically what we all learned in school, that the larger the optic disc, the larger the optic cup, and that can be completely normal. A classic example of Megalopapilla which you see commonly in most parts of the US and North America is in African-Americans. African-Americans tend to have a larger optic nerve, leading them to a larger optic cup. We know that demographically speaking, there is a higher risk of glaucoma in African-Americans than there is among the Caucasian population, for instance. We must also be careful, however, because if we are not somewhere like Chicago or Atlanta, if we are somewhere we are not used to seeing African-American patients, you may not have made yourself a normative database of the fact that a 0.5 or 0.6 cup is common and perfectly healthy within this ethic group. I think there are a number of African-Americans in which glaucoma is over diagnosed, and there are some for sure in whom we have missed it, particularly in practices that don’t dilate, take IOP, test visual fields, or take OCT readings of the nerve fiber layer and optic cup.

There are also patients for whom we have raised suspicions because, perhaps we don’t see that many African Americans, and we have a normative database based on Caucasian optic nerves and optic cups, in which a 0.3 C/D ratio is more normal for a person in their prime.

The patient in Figure 26 has Megalopapilla in their right eye, and we can see an oval cupping with the vessels on the nasal side as they merge into the disc. Figure 27, the patient’s left eye, gives us a closer view of the same patient, and we can see how we could easily mistake this for glaucoma if we had no baseline. Until the day comes that medical records reach back into childhood with digital pictures, or perhaps we do what I hear they do in Japan, where they take a picture of your optic nerve, but it on a thumbnail sticker, and put it on a card for you to carry around in your wallet. Thus, if you change locations or doctors, they can see what your optic nerve looked like on a certain date. Until we have that kind of thing, we are limited. Even then, a photograph is a two-dimension representation. We know now, those of us that are lucky enough to have access to scanning laser ophthalmoscopy, in the form of OCT or the Heidelberg Retinal Tomograph, we know that the C/D ratio from a 2D picture can be rather misleading.

Optic Pit

Another pseudoglaucoma that may occur at the optic nerve is an optic pit. Figure 28 is one that is associated with retinoschisis. When you have a splitting of the retinal layers,
you should always look for an optic pit, and vice-versa. It’s very critical if you find an easy-to-identify optic pit that you look for retinoschisis because that can cause vision loss, unlike the pit which isn’t progressive in any way. This one isn’t the easiest one to see. There is a macular hole in this image, and the patient will have reduced visual acuity if that doesn’t heal up well, and the schisis is all around the outer areas, which can lead to an off-macula detachment, a very dangerous, sight-threatening condition.

There is subretinal fluid often that happens with retinoschisis and optic pits, which can lead to the macular hole. We know in collie dogs there is active flow of fluid from the vitreous through the pit into the sub-retinal space. We have not yet demonstrated if this happens in people, but in Lassie-type dogs, this is the case. This is something to watch for, and is of particular interest to the surgeons who are looking to prevent the problem from occurring when it’s detected early. You can bet there are surgeons who are trying anti-VEGF injections of Lucentis or Avastin for this condition.

**Optic Pit and Disc Coloboma**

Sometimes optic pits will occur with coloboma, and this is an interesting topic. We don’t know where optic pits come from – we understand colobomas better. It’s certainly logical, and some authors have printed, that optic pit is a very, very mild coloboma. They have seen rarely the two occurring in patients, where they have a full-sized, or normal-sized, coloboma, and then a mini-coloboma in the form of an optic pit, so the theory goes.

There are some problems with this theory – the pits don’t occur where the fetal fissure occurs like the colobomas do. They often occur away from the fetal fissure. Patients often have pits on a unilateral, sporadic, unassociated basis, whereas colobomae happen usually with syndromes like Axenfeld-Rieger like we’ve seen before, and with systemic problems. Most often, pits are not seen in conjunction with retinal colobomae – they are usually seen in isolation. This theory may be logical, but may not be true.

The Greek word Koloboma, with a ‘K’, means defect. Colobomas are a hole in one or more structures of the eye; you can have an iris coloboma, a retinal or choroidal coloboma, or an optic disc coloboma. Here in Figure 31 we have a rare case where the pit is in the optic nerve, as always, but the coloboma is just off of the disc, so this is another one of those rare cases where the pit and coloboma occur together. Don’t expect to see this most of the time. However, in this case, we can see from the retinal vessels in this
picture that the optic cup looks anomalous and large, maybe a 0.8 C/D ratio or more. We would want to run an OCT on this one for sure. This would not be a progressive condition, but it may well affect the visual field and vision in that eye.

If you have a moderate coloboma, such as in Figure 32, it may take out most of the disc as in the picture. We can see the remnants of the disc at 12:00 or 1:00 or so, at the top of the figure, and it looks like there’s not much of a cup in that disc, as opposed to the easily visible cup in Figures 30 and 31. This coloboma is rather large, and adjacent to where the fetal fissure was – inferior to the disc. What we are seeing with all the white is sclera, not sensory retina. We will also have an enlarged blind spot here. And of course, we want to make sure we look for systemic abnormalities.

You want to differentially diagnose the coloboma above from a Myopic Staphyloma (Fig 33), of which I could show you lots of pictures. A myopic staphyloma is basically a scleral crescent, becomes a scleral ring, becomes a full-fledged staphyloma, which is different than a coloboma. In both cases you are seeing sclera, but with staphylomae we do not see this from birth. This happens, unless the patient is born with a double-digit myopia in the eye, this is happening as the axial length elongates. Essentially we are getting a stretch-mark ring around the optic nerve where we’ve lost sensory retina.

These patients may be at risk for retinal detachment due to the thinning of the retina with the elongation of the globe, or if they have Stickler Syndrome (a topic for another day). This particular one in Fig 33 is a pseudoglaucoma. It looks like there is something wrong, and there is, but it is a staphyloma, not a glaucoma. We are really looking at ectasia around the nerve.

This patient may have reduced vision, and we hope they don’t have this in both eyes. This tends to run in families, and would be a good indication for topical atropine during the growing years of many children, with informed consent of the parents, to see if you can decrease the elongation of the globe. I have tried this with kids using 0.125% Atropine, which does not sting and does not seem to cause significant cycloplegia for school or mydriasis for the sun.

**Abnormal nerves and nonprogressive field loss**

There are some other causes of pseudoglaucoma. Figure 34 shows a child who was up for adoption at the time. Left esotropia is pretty prominent, but if you’ve been in business for a while, you may also notice that this child also has fetal alcohol spectrum disorder. We can tell this by the missing groove between the nose and the upper lip, the thin upper lip, low-set ears, eso-tropia and strabismus in general is a higher risk. Notice the wide epicanthal folds for this kid’s age. He is in a crib, but that may be developmental delays as much as anything. If the
esotropia is not alternating, he should be patched for strabismic amblyopia and perhaps get surgery for the strabismus. There may be a Microcornea leading to a hypoplastic disc, which gives you a pseudoglaucoma that is not progressive because it is not a true glaucoma.

When they are not hypoplastic, the disc may be malinserted, as well. Malinsertion is different than tilting. We refer to malinsertion when the surface of the optic nerve is not parallel to the rest of the retina, whereas a tilted disc in optometry these days is referred to as a rotated disc relative to 12:00.

Figure 35 is a small, shallow optic nerve which, by itself, I don’t think anyone would call this a glaucomatous optic nerve. The vasculature is rather anomalous, but just over on the temporal side of this left eye, probably encroaching on the macula for sure, if not the fovea, is a giant coloboma.

This distracts you from what is going on in the eye. Visual field testing would show a field loss, but it’s not glaucoma that you would see here, as it’s not progressive.

**Optic Nerve Hypoplasia**

Hypoplastic disc may also be overwhelmed by tortuous vessels, as seen in Figure 36. There is a small, pale spot underneath that arrow, but notice the very small size of the disc. This is very common, and you will notice it also with fetal alcohol spectrum disorders, even those without the facial anomalies. There is Double Ring Sign that may occur in a hypoplastic disc, where there is a light ring around the outside of it, which we can see just a little of immediately to the right of the arrow in Fig 36, where the full-sized disc should have been.

There should be arcuate defects on visual fields if you are able to get a reliable one. The vision is down in this eye, and you are really looking at a pseudoglaucoma, or a glaucoma-mimicker.

**Malinserted Disc**

With a malinserted disc, we basically have the inferior portion on the right is a little bit elevated compared to the superior portion up at 10:00. This may also cause a crescent-like visual field defect. There is regional fundus ectasia here, and patients often end up with myopic astigmatism, with the power meridian oriented parallel to the ectasia. You can see this disc in Figure 37 looks a little bit elongated in the 2:00 to 8:00 meridian. The power meridian or the axis meridian is going at about 110 degrees or so.
You need to be aware of tilted disc syndrome because this may present with bitemporal hemianopia and optic disc elevation. If the nasal portion of the disc is to our upper left in Fig 37, it may not be seeing well due to the elevation. We may be worried in this case about the bitemporal hemianopia, optic disc elevation, which together sound like a pituitary adenoma or something at the chiasm. That will be non-progressive as well, so you have to be familiar with this condition.

In a child with malinserted discs, I would not order a CT scan just for this condition, but if they happen to have a CT scan for another reason, such as a bump to the head or other condition, it will often show anomalous nerves. You will see the nasal aspect of the globe is protruding posteriorly, as seen in Fig 38, and a field defect may result from this. Also, the optic nerves look abnormal, as well.

Figure 39 is the Goldmann Dynamic Manual Visual on the right eye – still done in some of these cases. You can see with the malinserted disc there is a superior temporal visual field defect that doesn’t quite respect the vertical.

**Morning Glory Disc**

I’m going to end with this part of the course by talking about Morning Glory Disc, and we will end the course by discussing the difference between that and Staphyloma and such.

Looking like the morning glory flower I showed at the beginning of this presentation, we are looking at a pigmentary disturbance and a retinal vasculature disturbance in which there is a sort of glial ‘bouquet’ or ‘tuft’ overlying the disc. It’s hard to see in a 2D picture. This is from a 1987 article, *The Morning Glory Disc Anomaly Contractile Movement Classification in Embryogenesis*.

Patients with Morning Glory Disc can see as well as 20/20 with this. However, often they have reduced acuities – 20/200 is not unusual. Sometimes the rather imprecise finger counting acuities or no light perception (NLP). These are unilateral conditions at least 80% of the time. There are some bilateral cases, but usually the other eye is doing OK, leading you to wonder whether this is some kind of acquired condition.
The patient may well have some acuity that is due to a deprivation amblyopia, if you will. We are used to defining amblyopia as a structurally normal eye that has some kind of interruption of development between 6 months and 2 years of age. This eye is certainly not structurally normal with the Morning Glory Disc, but sometimes a 20/200 eye, with some patching of the sound eye, can move down to 20/70. I usually look for one octave improvement, or a doubling of the acuity from the best corrected untreated acuities in these patients. Just because you know there is a Morning Glory Disc, that doesn’t mean you should not attempt patching, particularly a bandage patch in early childhood when the patient would not be in school yet. It certainly would only be done for only a couple of hours each day, and with parental supervision. You have to be careful once these kids are past infancy and they can crawl and move around, because you don’t want them to get hurt because they can only see 20/200. The idea is to try to improve the acuity whatever you can, so there is a secondary amblyopia, if you will, if such a thing can be said.

If you were to get a CT scan on these patients (Fig 41), you would see the funnel-shaped disc is showing up pretty well on a CT, even on a low resolution CT. It looks a bit like an optic neuropathy. You will see this more likely in African American and female patients.

We will now finish by doing some comparisons of optic disc coloboma to the morning glory disc.

Looking at Tables 6-2 and 6-3 from the Wright text (pg 223), let me point out a few things that I can call your attention to.

**Disc coloboma** is your main differential diagnosis from a Morning Glory Disc. If you see that the disc is contained centrally within the excavation as it says, consider Morning Glory. Look for that tuft or bouquet of glial tissue, pigment disturbance, and anomalous vasculature. If you don’t see any of those things, and they all look normal, it could be that you’re looking instead at an optic disc coloboma.

Further comparisons between the two conditions – you should see other
associated colobomas, often in both eyes. You should see colobomas in the family, and systemic syndromes in these patients, such as Axenfeld-Rieger in a patient with optic disc coloboma. None of that should happen in a patient with Morning Glory Disc.

If you look instead for a staphyloma instead of a coloboma to differentially diagnose from Morning Glory Disc Anomaly, (Table 6-4, Wright 3, pg 224) you are going to have a good optic disc with normal vessels in peripapillary staphyloma. You also will not see pigment around most of the time around a staphyloma, but I will show you one exception here in a moment. The excavation in a staphyloma will be deeper than that of the Morning Glory Disc Anomaly.

Figure 42 is an example of a peripapillary staphyloma that looks a lot like a Morning Glory Disc to me. This is a normal disc, the cupping looks OK from the vasculature contour that we can see in a 2D picture. We know this is not a Morning Glory Disc because the vessels look normal, there is no central bouquet of glial tissue, especially if you had a 3D slit lamp view. We would also be able to see that the depth to this is greater than that of Morning Glory Disc.

**Take-Home Pearls**

Some things to study for your test, if you are going to be taking it for credit:

1. Many secondary glaucomas are due to congenital causes
2. Most childhood glaucomas are secondary to anterior chamber dysgenesis
3. Secondary glaucomas are usually treated surgically by a glaucoma specialist
4. Some are accompanied by posterior segment anomalies
5. These congenital posterior segment anomalies can mimic glaucoma in structure and in function (visual fields)

The secondary glaucomas, the true glaucomas, which happen in children are due to anterior segment abnormalities that are mostly congenital. They can happen later in life; you are born with the unusual anterior segment, and it gets you glaucoma later, for example at age 20. So be aware of that – this goes beyond traditional pediatric age.

The anterior chamber is usually not completely formed. Maybe the tunica vasculosa lentis did not completely recede. You need a surgery and maybe a glaucoma specialist to treat these, but afterwards if they stop seeing the surgeon and come to see you for co-management, you will need to be aware of them.
The posterior segment abnormalities do go with some of these conditions, particularly the colobomae, but there are other ones that look like glaucoma because they have field loss and the optic nerve looks anomalous. Those ones you need to be aware of.

If you need to get ahold of me, below is my contact information. I run our 3D vision clinic in Beaverton, OR. As I said before, I do pediatrics for sure, and I teach Visual Perception, Ocular Motility, Nutritional Optometry and Pediatric Ocular Disease, from which this lecture has risen.

I am happy to hear from you via email. Thanks for supporting Pacific University’s Web CE program, and I look forward to hearing from you in the future.

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