

“Chemical Eric” Can’t See

by
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The Eye Exam

It was a sunny day in 2005, and time for Eric’s annual eye exam. Dr. Tim flipped through his charts.

“You know, we really should measure your visual fields. It’s been at least ten years since you had that done!”

“I hate them!” Eric grumbled. “I get disoriented and, anyhow, we already know I have a large blind spot.” He raised his hand, squinting through one eye. “Here. I can’t see my finger here.”

“I know.” Dr. Tim was sympathetic. “But weren’t you just telling me you haven’t seen stars for a while? It’s important. You had that tumor on your pituitary gland. Your optic nerves cross in that area, right in front of the pituitary, so if anything is happening again we’d probably notice it first by changes in your eyesight. It really is important!”

An hour later, Eric was seated in front of a large white screen, staring intently at a light in the middle. “Keep looking at that light, so your eyes don’t move. If you see another light anywhere else, push this button.”

Eric began the test by focusing on the light he saw in the center. The machine then successively displayed points of light of varying intensity in different places around the entire visual field. When Eric saw the peripheral light, he pushed a button. He did his best to detect the flashes, but realized he was probably missing some. The machine mapped out where he could see and where he could not.

Dr. Tim looked at the printout. His face grew serious. “Eric, we have a problem. You have limited peripheral vision, and it’s bad enough that I don’t think you should be driving! This looks to me like something called retinitis pigmentosa. We should refer you to a specialist.”

He pulled out more papers. “Here, look at this. Here are your visual fields. The numbers show how well you can see in that area of your eye, and in the graph on the right the darker the box is the less you can see. Now here are a normal person’s visual fields. Do you see the difference?”



Questions

1. How do your eyes work? What is the difference between focal vision and peripheral vision?
2. What is the anatomy around the pituitary gland? Why are the optic nerves likely to be affected by pituitary tumors?
3. Interpret the results of Eric’s visual field tests (shown on the following pages). How does the visual fields machine map peripheral vision, and what do the numbers represent? What is the dark area in the “normal” eyes? Based on these visual fields, is Dr. Tim correct in suggesting that Eric should no longer drive?

Normal Left Eye

Single Field Analysis

Eye: Left

Name	DOB:
ID:	

Central 30-2 Threshold Test

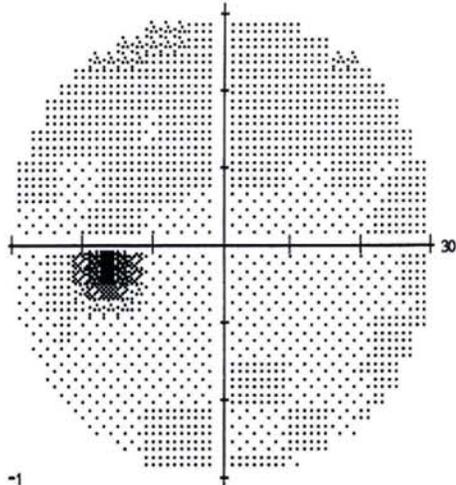
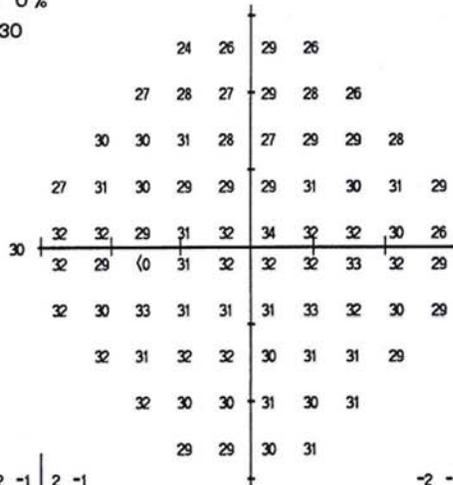
Fixation Monitor: Gaze/Blind Spot
 Fixation Target: Central
 Fixation Losses: 2/16
 False POS Errors: 4 %
 False NEG Errors: 0 %
 Test Duration: 06:30

Stimulus: Ill. White
 Background: 31.5 ASB
 Strategy: SITA-Standard

Pupil Diameter:
 Visual Acuity:
 RX: +0.00 DS DC X

Date:
 Time: 5:19 PM
 Age:

Fovea: OFF



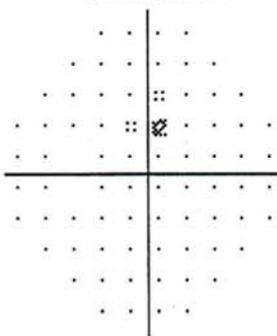
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-1	-1	-2	0	-1	-3				
0	0	1	-3	-4	-2	-1	-1		
-3	1	-1	-3	-3	-4	-2	-1	0	0
1	1	-2	-2	0	-2	0	-1	-3	
2	-3	-2	-2	-2	-2	0	1	0	
1	-1	1	-2	-2	-2	0	-1	0	0
1	0	0	0	-2	-1	0	0		
1	-1	-1	1	0	1				
-1	0	1	2						

-2	-1	1	-1						
-1	-1	-3	0	-2	-4				
0	-1	0	-3	-5	-2	-2	-2		
-4	0	-2	-3	-4	-5	-2	-2	0	0
1	0	-2	-2	0	-2	-1	-2	-4	
1	-3	-3	-3	-3	-2	0	0	0	
0	-1	0	-2	-2	-3	-1	-1	-1	0
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0	-1	-1	0	-1	1				
-1	-1	0	2						

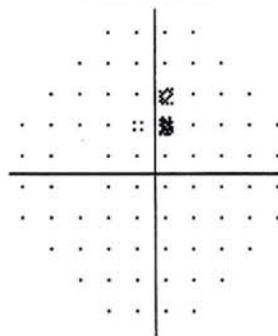
GHT
 Within normal limits

VFI 99%
 MD -1.00 dB
 PSD 1.62 dB

Total Deviation

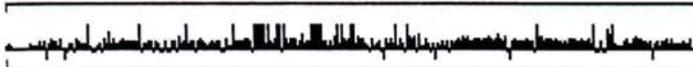


Pattern Deviation



:: < 5%
 ☼ < 2%
 ☼ < 1%
 ■ < 0.5%

Professional Eyecare Center
 Dr. Tim Bengtson
 820 E. Jackson
 Macomb, IL 61455
 (309) 833-4242

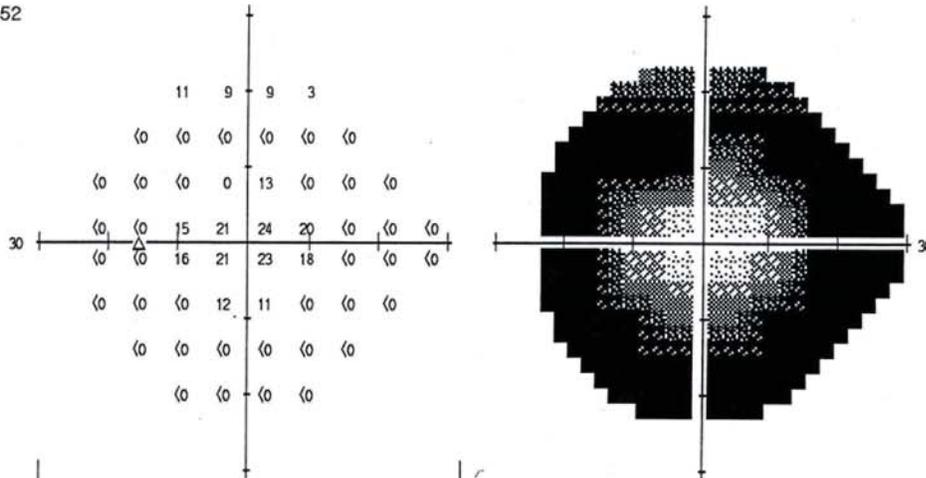


Eric's Left Eye

Single Field Analysis Eye: Left
 Name: Ribbens, Eric ID: DOB: 12-19-1957
 Central 24-2 Threshold Test

Fixation Monitor: Gaze/Blindspot Stimulus: III, White Pupil Diameter: Date: 06-01-2005
 Fixation Target: Central Background: 31.5 ASB Visual Acuity: Time: 7:10 AM
 Fixation Losses: 0/14 Strategy: SITA-Fast RX: +1.50 DS DC X Age: 47
 False POS Errors: 0 %
 False NEG Errors: 0 %
 Test Duration: 05:52

Fovea: OFF



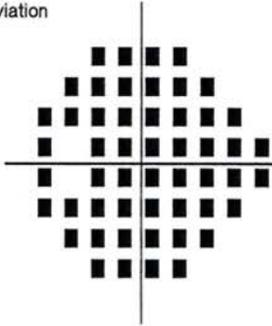
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-31	-32	-32	-32	-32	-32		
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-33	-17	-12	-10	-15	-34	-32	-30
-33	-33	-34	-21	-22	-35	-34	-32
-33	-33	-34	-34	-33	-32		
-32	-32	-32	-32				

0	-2	-3	-9				
-15	-15	-16	-16	-16	-15		
-15	-16	-17	-15	-3	-17	-17	-15
-16	-1	4	7	4	-18	-16	-14
-16	0	4	6	2	-18	-16	-14
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-17	-17	-17	-17	-17	-16		
-16	-16	-16	-15				

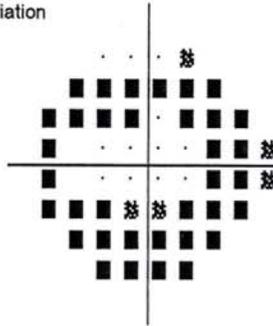
GHT
Outside normal limits

MD -27.54 dB P < 0.5%
PSD 8.66 dB P < 0.5%

Total Deviation

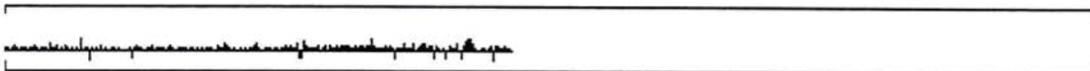


Pattern Deviation



∴ < 5%
 ☼ < 2%
 ☼ < 1%
 ■ < 0.5%

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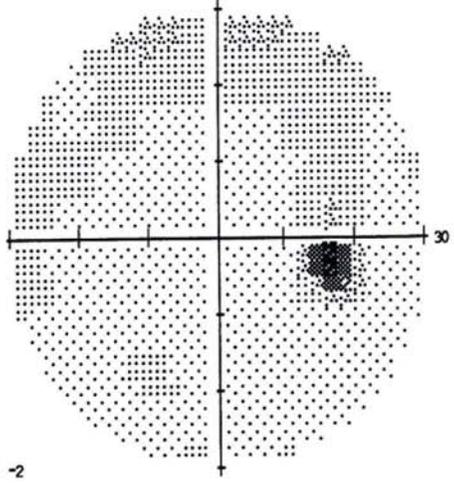
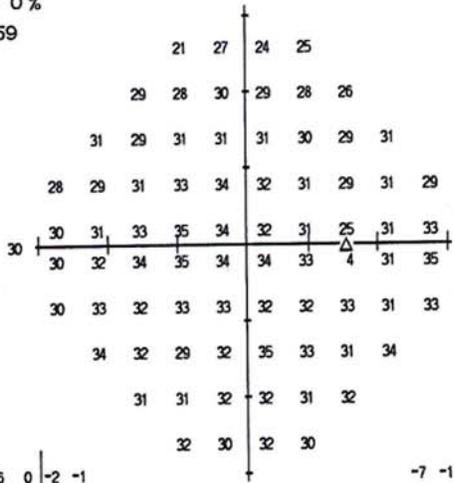
Normal Right Eye

Single Field Analysis Eye: Right
 Name: _____ DOB: _____
 ID: _____

Central 30-2 Threshold Test

Fixation Monitor: Gaze/Blind Spot Stimulus: III, White Pupil Diameter: _____ Date: _____
 Fixation Target: Central Background: 31.5 ASB Visual Acuity: _____ Time: 5:07 PM
 Fixation Losses: 1/16 Strategy: SITA-Standard RX: +0.00 DS DC X Age: _____
 False POS Errors: 7 %
 False NEG Errors: 0 %
 Test Duration: 05:59

Fovea: OFF



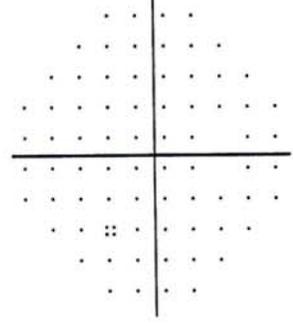
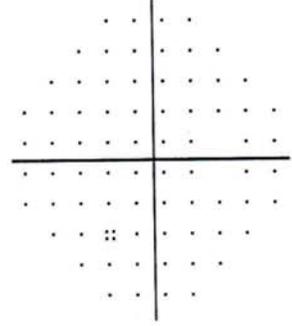
-6	0	-2	-1
0	-1	1	0
1	-2	0	0
0	-2	-1	1
1	0	0	1
1	1	1	1
1	2	0	0
4	1	-3	0
2	1	1	1
3	1	3	0

-7	-1	-3	-2
-1	-2	0	-1
0	-3	-1	-1
-1	-3	-2	0
0	-1	-1	0
0	0	0	0
0	1	-1	-1
3	0	-4	-1
1	0	0	0
2	0	2	-1

GHT
 Within normal limits
 VFI 100%
 MD +0.15 dB
 PSD 1.58 dB

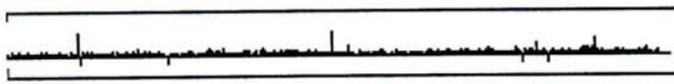
Total Deviation

Pattern Deviation



∴ < 5%
 ◐ < 2%
 ◑ < 1%
 ■ < 0.5%

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Attitudes to Genetic Diseases and Disabilities

Genetic diseases are caused by mutations in the gene associated with the trait. These mutations can be dominant (always expressed), recessive (only expressed if both chromosomes have the mutated gene), or as increasing the probability of acquiring the disease (e.g., many forms of cancer).

Questions

1. Do you know someone with a genetic disease? Do you know someone with a disability? If you do, how do you think their problem has affected them?
2. Imagine someone with an obvious disability; for example, someone in a wheelchair. Do you treat someone like that differently than “normal” people? Do other people treat someone in a wheelchair differently? How?
3. Imagine your doctor told you that you had a genetic disease, such as Eric was told. Further imagine that you are told there is nothing that can be done about it. How would you feel? What would you do?
4. Predict what the future holds for Eric.

The Retina Specialist

Dr. Pau folded his hands. “I’m afraid you do have retinitis pigmentosa. It’s really a syndrome, with more than 70 known genetic mutations that can cause it, but in all cases the pigments lining your eye gradually break down, so you can’t sense the light that hits your eye.”

Eric winced. “Great. So what do we do about it?”

Dr. Pau shook his head. “Not much, I’m afraid. One study has shown that taking vitamin A may slow the rate of loss, but the loss is irreversible. Typically, patients slowly lose their peripheral eyesight, at a rate of say 5% loss per year. The speed can vary enormously, and in many people it can slow or simply stop. But in other cases patients become blind. The good news is that it usually starts in adolescence or early adulthood, but you are in your late 40s. Oh, and wear sunglasses whenever you are outside.”

Dr. Pau continued: “You probably have a version of retinitis pigmentosa named Usher’s Syndrome. It usually has both hearing and vision loss, and is named after C.H. Usher, a British ophthalmologist who described the syndrome in 1914. Ten to fifteen thousand people in the United States have Usher’s Syndrome. It is apparently the most common cause of deafness with blindness. There are three types, and apparently Type I is the most common.”

Table 1. Types of Usher’s Syndrome

Type	Hearing	Balance	Vision
Type I	Complete deafness at birth	Problems with balance	retinitis pigmentosa
Type II	Moderate to severe hearing loss at birth	No balance problems	retinitis pigmentosa
Type III	Progressive hearing loss	No balance problems	retinitis pigmentosa

“I’ve always had some hearing loss,” Eric remarked. “In fact, I used to wear hearing aids, but they were a real nuisance, so I threw them away. But it’s always stayed the same, so I probably have Type II.”

“Well, I wish I could do more to help you. You already are compensating by doing things like turning your head. They are working on a clinical test of a gene therapy for macular degeneration, so maybe in 10 or 20 years they will have a way to treat retinitis pigmentosa.”

Questions

1. How could a genetic mutation cause something like this to happen?
2. How could a genetic therapy be used to treat an eye problem?
3. Why aren’t doctors sure that taking vitamin A supplements slows the rate of progression? What evidence would you need to be sure of this? If they aren’t sure, should Eric be taking vitamin A anyhow?
4. Dr. Pau said this was a genetic disease. How would you determine whether the condition is recessive or dominant?
5. Imagine that there is a genetic therapy. They stick a needle into your eye and inject the working gene, which then hopefully binds to the cells in the retina that should be making these pigments. Would you do it? What if it would protect your remaining eyesight, but wouldn’t restore what’s left?
6. What do you think are Eric’s concerns at this point?
7. Eric was attending a conference and stayed in a hotel. When he left, walking through the lobby with suitcases and briefcases loading him down, he did not see that someone had placed a low “coffee table” in the middle of the foyer. Eric walked right into it and fell over it, bags tumbling. Embarrassed, he staggered up and picked up the scattered luggage. The clerk behind the counter cheerfully said, “Oops! It’s like you were blind or something!” Eric muttered something and limped outside, sore and mad at himself. Why would Eric feel embarrassed about something that isn’t his fault?

Trying to Visualize Retinitis Pigmentosa

It's hard to imagine what no peripheral vision is like because if you have no peripheral vision you instinctively want to look to the side, and then of course whatever you are looking at is in focus. One way to imagine it is to build a cardboard "binoculars" with two toilet paper cores. Tape two cores together and navigate the room while looking through them. If you want to see something you have to be constantly turning your head to focus on what you want to see.

The image at <http://www.aoa.org/Images/retinitis.jpg> is an attempt to show what the effects of retinitis pigmentosa are like. It's really not a very accurate image, because it's not at all what you really see. You don't see the ring of black or anything like that. It's not black; it's just the absence of side vision. Think about what you see beyond the edges of your vision. It's more like cropping a photograph. For Eric it's not so much black around the margins as fuzzy, with a halo of gold sparkles at the edges of his vision.

Two helpful videos:

- Retinitis Pigmentosa: What We Can See. http://www.youtube.com/watch?v=fAf1_kbt5cw.
- Rikard André: Retinitis Pigmentosa at Night. <http://www.youtube.com/watch?v=eggmXiFxsEo>.

Vitamin Analysis

There is no way to reverse either the hearing loss or the vision loss of retinitis pigmentosa. There is some evidence that taking vitamin A supplements will slow the progression of Usher's Syndrome Type II vision loss and the loss in typical retinitis pigmentosa (RP). The evidence is from a study published in 1993 that tracked adults with RP for six years. It concluded that taking 15,000 international units of vitamin A palmitate daily slowed vision loss by about 20%. Note that the volunteers were adults with RP and Usher's Type II, so these findings cannot be extrapolated to children or individuals with other forms of Usher's Syndrome. Conversely, vitamin E doses of 400 IU or higher may increase the rate of progression.

Questions

1. If the normal rate of loss is to lose about 5% of your remaining vision each year, and if the study above is right that vitamin A slows that rate 20%, compare someone with 50% vision loss taking vitamin A to someone with 50% vision loss who does not take vitamin A. How many years would it take for these two individuals to lose 90% of their vision?
2. What is vitamin A? Why don't scientists know for sure whether vitamin A helps?
3. Try to reconstruct the research design of the vitamin A study. Given only what you are told above, what can you discern about what they did? Are there other ways to investigate this question?

Coping with a Disability

Eric has a serious disability. He is legally blind, no longer drives, and cannot see the entire screen of a computer anymore. Place yourself in the following (true) scenarios. What would you feel if you were Eric? What would you do?

Scenario A: Eric is reading anonymous student comments in his course evaluation. One student wrote: “He can’t see! He should get his eyes fixed!”

Scenario B: Eric is walking through a crowded department store. Because he is so tall, he doesn’t see short people who are close to him unless he is looking almost straight down. He doesn’t see a woman coming, and bumps into her hard. She gasps, “Well! You clumsy ox!”

Scenario C: Eric is playing wiffle ball with his daughter. When she throws it at him he can’t follow its path and he flinches. She shouts “Come on Daddy! Catch the ball!”

Scenario D: Eric is sitting in a restaurant, talking to a professional acquaintance. Animatedly he gestures and a large water glass he didn’t notice gets knocked over, spilling cold ice water all over his tablemate.

Scenario E: Eric is working on the computer and he loses his mouse cursor. It is somewhere on the screen, but he doesn’t see it.

Scenario F: For years before Eric was diagnosed with retinitis pigmentosa, he couldn’t see stars and his wife would remark he was a reckless driver. So his disease must have been affecting his sight for years before he acknowledged it. Why do you think Eric ignored signs that he was developing a vision problem?

Scenario G: Recently, Eric has started wearing an eye patch. Now he has an obvious sign of a disability. How do you think people treat him differently?



Genetic Inheritance Patterns

Retinitis pigmentosa (RP) can be autosomal recessive, autosomal dominant, or x-linked. Apparently the dominant forms are often less severe. Usher's Syndrome is an autosomal recessive inheritance (i.e., you must get a copy of the defective gene from your Mom and one from your Dad). Autosomal means it is not carried on one of the chromosomes that determines sex. One website (<http://www.emedicine.com/oph/topic704.htm>) says that Usher's Syndrome Type II has been mapped to chromosome arm 1qe. Usher's Type II is recessive, so for Eric this means that both his Mom and Dad are carriers of this condition.

Questions

1. Draw a pedigree of Eric's family showing possible genotypes and chances of having RP. Include his parents, Eric, his brother Dirk, and a daughter of Eric's.
2. Imagine you are a genetics counselor. What advice would you give Eric's brother Dirk about the chances of Dirk's children having retinitis pigmentosa? (*Hint*: assume that there is a 1:80 chance that Dirk's wife is a carrier.) How would your advice change if his wife has the disease?
3. Should Eric have children? Imagine he has a daughter (he has two, but both are adopted). What advice would you give a genetic daughter from a genetics counseling viewpoint?

A Poem

Below is a poem that Eric wrote about pigmentosa. At the time, he was coming to grips that he really had a vision problem.

Retinitis Pigmentosa

I lie in bed and gaze into the universe's soul through my skylight.
The refracted blue curtain has been pulled; her serenity the moon is visiting China.
The sky overflows with starlight traveling long decades for this moment.
But I cannot see the stars.

Shadow on shadow, shades of purple and gray. Dimly I see dark window edge.
And there, trembling on the edge of awareness...Yes, there!
Straining to fix her, triumphant, I find a star.
(It is probably Venus, but I am pretending.)

Oh, I know they are there. I remember that shimmering pale blaze,
Remember their gleam as an old man remembers long-lost lover's touch.
If you were here you would share your vision.
No, I will not shed tears for lost sight again.

I mumble my prayer: "I thank you, Lord, that I do not have macular degeneration."
My selfish depravity swells and I am ashamed.
A single tear slips to water my beard.
(Must be allergies.)

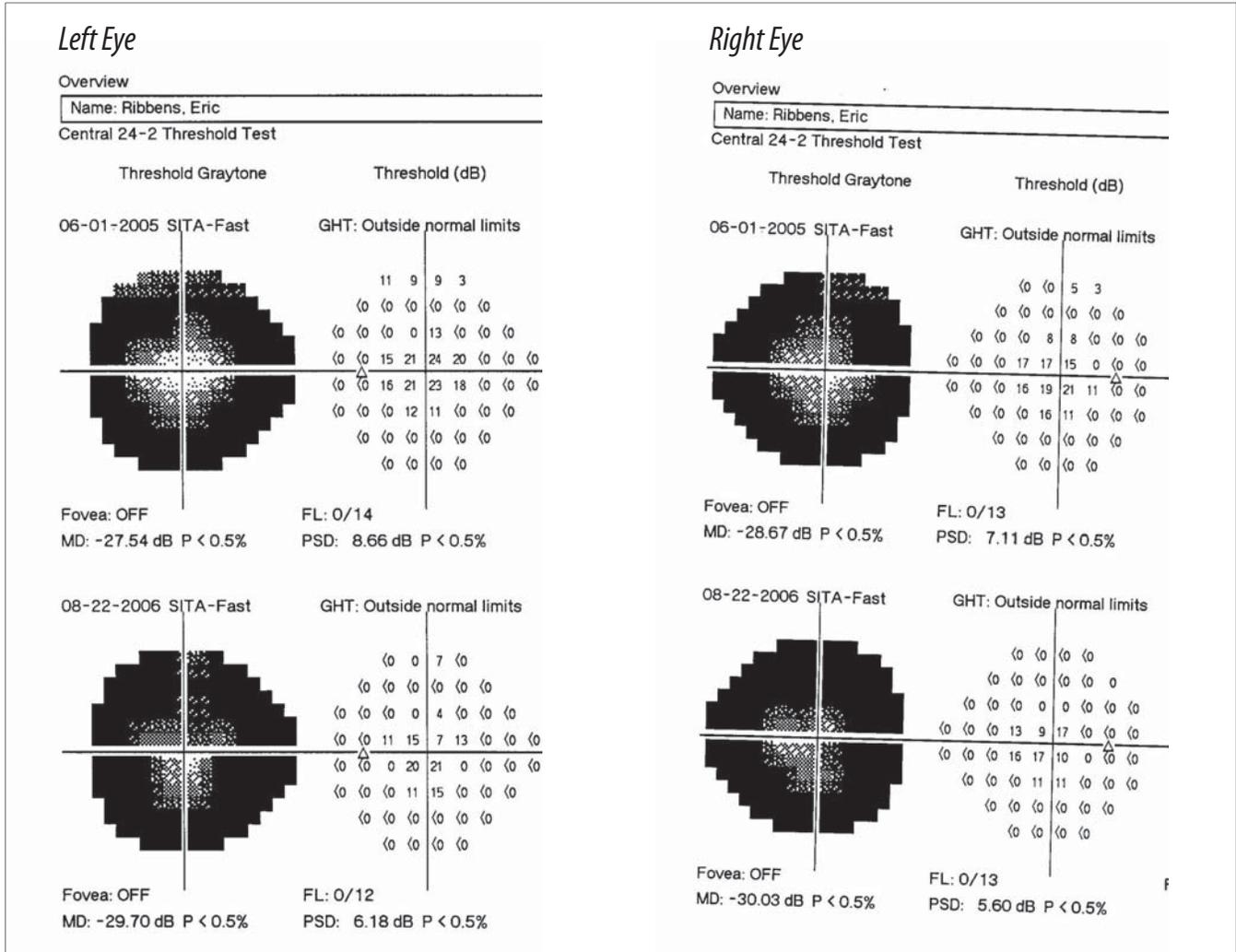
Questions

1. Why can't Eric see stars? How does he feel about that?
2. "If you were here you would share your vision." How can you share your vision with a visually impaired person?
3. He says he won't cry about lost sight again. Is this what he should do?
4. What is macular degeneration, and why is Eric glad he doesn't have it?
5. The last paragraph is clearly religious. Eric is a Christian. Can a scientist really be religious?



Deterioration Analysis

Fourteen months after Dr. Tim, Eric's optometrist, first diagnosed Eric with RP, he ran a set of specialized tests that measure the ability of different parts of the eye to see. The figures below show the results, compared to 14 months ago. Each image shows the visual acuity of the eye, and the numbers to the right of the image show how well Eric can see in each portion of his eye; left corresponds to left eye, right to right eye, and the top two figures are from 2005 and the lower two figures from 2006. Thus, the number 11 in the top line below means that in the top left of his eye he could see at an intensity of 11 fourteen months ago. Dr. Tim was excited, because the numbers could be compared directly to determine how rapidly Eric's eyes were deteriorating.



Questions

- Over the past 14 months, how much has Eric's left eye gotten worse? His right eye? *Hint:* there are two ways you could do this. You could calculate the percent of numbers greater than zero (a measure of the overall proportion of the visual field he can see). You could also measure the percent decrease in the ones in which he can still see (a measure of the quality of vision in the area he can see).
- After seeing the results of these tests, Eric stopped driving, gave his truck to his brother, and cancelled his car insurance. Was this a good idea? What else should he do?
- What should he tell his students about his eye disease at the beginning of the semester?

Postscript

I was diagnosed with retinitis pigmentosa and that year I hit two deer, bumped into a pair of students, and backed into a telephone pole. I decided that I wouldn't be able to forgive myself if I ever really hurt someone. I gave my truck to my brother Dirk, who promptly named it George. Today I get around by walking, using the university bus system, and by my wife driving me places. My eyesight has continued to degenerate, and I'm now legally blind, which means I have a focal area of less than ten degrees. I also wear an eye patch now because of damage to the 4th, 5th, and 6th nerves for my right eye. Going from bright to dark places is difficult, and I often bump into things or stumble over stuff. But I'm still teaching, and I study plants, which don't move!