Patients in the driving seat: Birdshot chorioretinopathy

Birdshot chorioretinopathy, usually known just as 'Birdshot', is a rare and little-understood condition. Early diagnosis is vital, but it can be difficult to identify. Radhika Holmström investigates.

"I didn't realise there was anything wrong with my eyes till the night I woke up and couldn't see at all out of one eye. Even then I didn't really worry. Birdshot tends to be very slow, very gradual and you do not realise you are losing your sight until it is nearly too late," says Annie Folkard.

Folkard and her colleague Rea Mattocks are the driving force behind the inaugural conference of Birdshot Uveitis Society on 11 September this year. The conference will bring together the relatively small number of people (estimated to be a maximum of 500 throughout the UK) who have a diagnosis of Birdshot Chorioretinopathy, as its full title is (it is usually shortened to Birdshot). It is also the launch of a patient-driven partnership with health professionals – with patients firmly taking the initiative and setting the parameters.

The condition

Birdshot is officially classified as a 'rare disease' – which means there are far fewer official rules about how it should be approached and treated. It is also a relatively new disease: "It's only really been understood since the 1980s," says Mattocks. Like conditions ranging from diabetes to rheumatoid arthritis, it is an auto-immune disease, caused by the body's immune system attacking its own tissues. "Uveitis encompasses many forms of inflammation in the eye. As with all inflammation, the immune system becomes disregulated and starts attacking the system rather than protecting it," explains Andrew Dick, professor of ophthalmology at Bristol University.

Specifically, Birdshot is a form of 'posterior uveitis', affecting the retina and the layer of tissue that supports it. It is not surprising, given that the retina is involved, that posterior uveitis tends to be more damaging than the other forms – and Birdshot can certainly lead to blindness if left unchecked.

At the moment, it's not clear why some people develop Birdshot chorioretinopathy. Most – though not all – people with the condition also carry the antigen HLA-A29. However, many people who test positive for HLA-A29 never develop Birdshot, so it seems clear that a 'trigger' of some kind is necessary to make the eye react to it in this way. "There is an awful lot being done on the mechanism that causes inflammation in the eye. We know a lot about how you damage the retina and the antigens that may be involved, and increasingly realising why and how people respond," Dick explains. "Birdshot is linked genetically with this antigen – but at the moment we do not know quite what that means."

Diagnosis

A lot of forms of posterior uveitis start with blurred vision, and/or floaters in the eye. Birdshot is no exception. People do experience other symptoms such as sensitivity to glare, decreasing peripheral vision, poor night vision and/or poor colour vision. For many, floaters are the main thing they notice, especially if their central vision remains relatively intact while the disease takes hold – and as floaters are very common, this symptom is often overlooked.

"People come to casualty and are sent away, and all the time their disease is ongoing. The inflammatory process will still be destroying the cells of the retina even if the central vision remains good. That is the other thing that throws off optometrists. Most uveitis will drop your central vision, and certainly Birdshot can too, but central vision acuity is not a very good marker for the disease," says Narcisse Okhravi of Moorfields Hospital. "You need to dilate the eye and examine the vitreous using the specialist 'slit lamp', in order to check for inflammatory cells. If you have identified those cells you know it is likely to be posterior uveitis, which is a good start. Then you've got to get the patient to a specialist uveitis clinic."

At a specialist clinic, such as the Moorfields one, the condition is detected by a full ocular examination, including angiography and electrodiagnostics. In a person who has had Birdshot for a while, there are usually oval ➜

Rea Mattocks: A personal view

Unlike many people with Birdshot, Rea Mattocks developed the condition virtually overnight. She describes her experience.

In another situation I would have gone blind by the time I got treatment, because the first registrar I saw just told me it was the ageing process. Because of my work, I have been able to insist all the way along on seeing a consultant. Even at that early stage he queried whether it was an auto-immune disease, and then identified that characteristic look on the retina.

There is a huge inequality in the treatments available for rare disease, and very blunt instruments for dealing with them. I couldn't tolerate the medication I was put on – it made me completely loopy, to the point where I thought I could fly, and didn't stop my sight deteriorating either. I had to give up work. My current steroid does seem to be holding this at bay. However, all the medications interact with each other. My bone density is low and I am on medication for that – and my cholesterol has hugely increased, so I am on statins for that. My blood pressure is up hugely too, so I need to exercise but at the same time have to be careful about that. I often wake up and vomit. My energy levels have plummeted. I have skin problems – I just have to touch it and I bruise or the skin tears – and ironically I have blepharitis, another eye condition. From being someone who had never taken a sick day, I am now on medication for the rest of my life.

I looked up everything I could and got in touch with the few experts. I flew across to the US for a small Birdshot conference in October 2008 and for the first time I met other people with it, including Annie Folkard whose experience had been so different from mine. We came back to the UK and wrote to every single uveitis consultant asking their views on our setting up a support group. Two thirds wrote back and we found 300 patients between them – but you can bet there are more out there.

shaped orange to cream spots (the 'birdshot') in the retina, marking the characteristic damage caused by this disease. Yet even at a centre of excellence like Moorfields, it can take up to 18 months to reach a definitive diagnosis, because the disease can operate in such different ways that it is not always easy to recognise.

Treatment

"Treatment is proven to work. It has even been shown that after two years of not controlling the inflammation, the patient will improve. And it's important to tackle it before the central vision acuity is attacked," Okhravi says. "Some people do recover vision they previously lost, but the most important thing is to stop it getting worse." The problem here – and it is a big one, as significant as getting a diagnosis in the first place – is that because every case varies so much, so does the treatment they need. In addition, some ophthalmologists still believe that this is a condition that will run its course and finish, so that their patients go untreated for even longer. There is already a "managed clinical network" in Scotland for people with Birdshot – a clinical pathway, with a clear treatment protocol – but at the moment, this is lacking in the rest of the UK.

"All the medications interact with each other, and you need massive dosages. But you either go on this toxic regime, or you go blind"

The treatment usually targets the inflammation using steroids, and then uses immuno-suppressants to stop the inflammation attacking the eye. One of the latest theories is that patients need two forms of immuno-suppressant to tackle different aspects of the condition. These are all very strong drugs, and often produce side-effects which then in turn need to be tackled with their own medication, as Mattocks points out. "All the medications interact with each other, and you need massive dosages. But you either go on this toxic regime, or you go blind." It's commonly recommended that people receiving treatment for Birdshot have their liver and kidney functions tested, along with their blood pressure: and, if they have been on oral steroids for some months, they will need their bone density checked as well.

BUS: the support group

With so little known about the disease, patients are taking control of their agenda – rather as they have done with the Alström Syndrome UK Support Group (see NB, October 2009), which Mattocks and Folkard both took as a model when setting up the Birdshot Uveitis Society, a support group for people affected by the condition.

"We know that Birdshot patients in the UK are very isolated. We need to start by establishing how many people we know have the condition, and getting health professionals to realise that there are probably others going undiagnosed. We are getting together a committee made up of the full 'Birdshot community': ophthalmologists, uveitis nurses, specalists in rare diseases and so on," Mattocks explains. They hope that the launch day will lead to the start of a wider protocol for managing and treating Birdshot and to an agreed programme of research.

"We're hoping that we can find all the patients in the UK, and educate them so that they can get as much help as possible. Then we want to raise the awareness of the disease among anyone who may be dealing with patients with floaters: getting them to ask a few questions and look at the vitreous," adds Okhravi. "And we also need to set up more research, which is

needed at every stage. We need better identification, better tests more readily available in the clinic, and better treatments with fewer side effects."

Dick agrees about the potential for finding treatments. "I think this should make it possible to slow down the progression of the disease. Now we have the patients in this number we can get a much clearer picture of how the disease operates, get a clearer picture of the patients' genes and interrogate the potential antigen they are responding to. That may mean we can introduce tolerance therapy, which re-educates the immune system not to react to that antigen." Tolerance therapy has reached phase two trials with multiple sclerosis and diabetes, he points out.

Whatever next step the specialists take, however, they are clear that the patients themselves will determine it. "Whatever we do, we need to ensure that everyone is in this together. We need patients to tell us their concerns, about the treatments they have had and the difficulties they are having – and the research they think should be done," says Okhravi.

Dick concludes: "The excitement with patient groups like this is that it is really power to them. We are essentially here to serve. I love this – it is exactly how it should be, with patients dictating what they want and saying what they want, on their own terms."

Further information

The Birdshort Patient day is on Saturday 11 September, 10am to 4.45pm, at UCL Roberts Building, Torrington Place, London WC1.

Information and tickets are available from Birdshot@live.co.uk or by post: BUS, PO Box 64996, London SW20 2BL or birdshot.org.uk