There are two categories of conditions of the retina: retinitis pigmentosa (RP) or rod-cone dystrophy, and macular degeneration or cone-rod dystrophy. Retinitis pigmentosa includes diseases of the outer part of the retina, the part where the rods are located. Remember that the rods are the photoreceptors that capture images of movement and are active in dimly lit situations. Macular degeneration, on the other hand, is a degeneration of the cones, our central vision which is used for reading, seeing colors and looking at details in well-lit situations.

Remember Jonathan
You met Jonathan in Unit 1. He’s an eighth grader who has lost his side vision. He bumps into people and objects when he’s tired, and he looks lost when he enters the school auditorium and the lights are already dimmed. Over the years his vision has changed. He does his schoolwork in braille, but he also reads magnified print when necessary. His orientation and mobility teacher has given him a series of evening lessons to impress upon him the need to use a cane in dimly lit situations. Jonathan has been diagnosed with Leber’s congenital amaurosis, a form of retinitis pigmentosa.

The residual vision for people who have retinitis pigmentosa is referred to as “tunnel vision” because it is only in a narrow, central part of the eye. This type of RP is also referred to as rod-cone dystrophy.

Often when people have retinitis pigmentosa, they first notice that their vision is poorer at night. In fact retinitis pigmentosa is sometimes called “night blindness.” People who have it don’t notice obstacles in dim lighting, and even in well lit areas they may bump into things which are not directly in front of them. They lose some of their contrast sensitivity, making it difficult to see subtle changes in terrain such as cracks in the sidewalk or even curbs. Many people who have retinitis pigmentosa lose all of their vision gradually, and some retain central vision.

Myopia (nearsightedness) often accompanies retinitis pigmentosa. Large print is often useful for readers who have retinitis pigmentosa, but care must be taken not to enlarge letters and symbols outside of the visual field. Braille is usually a preferred medium for reading and writing as vision loss progresses.

Meet Cam
Cam was just diagnosed with Stargardt’s disease last year at the age of eight. He was becoming more and more frustrated in school because of difficulty reading. His teachers and family members didn’t notice his vision loss at first, because Cam continued to run around just as he always had. At the school vision
screening Cam’s acuity loss was noticed, and in follow-up visits to his private physician, Stargardt’s was eventually diagnosed. Now Cam is catching up with his classmates, using magnification for reading. He is also beginning to learn braille.

When a condition causes the degeneration of the cones first, it is called macular degeneration or cone-rod dystrophy. There is a loss of detail and color vision. People who have cone-rod dystrophies rely on large print and, more frequently, braille for reading and writing.

There are many syndromes which include RP as the visual condition. Some of them are Bardet-Biedl and Lawrence-Moon-Biedl, Leber’s congenital amaurosis and Usher’s syndrome. Each syndrome has distinct features and a different pace and extent of vision loss. The most well known type of macular degeneration is age-related macular degeneration. This occurs in many elderly people, but you will not see it in school children. Stargardt’s is the name given to the type of macular degeneration seen in children. Alstrom’s syndrome and Batten’s disease are two more conditions which include cone-rod dystrophy.

Here are descriptions of some of the syndromes which occur in children and include RP or macular degeneration. The first three all have vision loss that progresses from the outer periphery vision toward the central vision. The others are marked by central vision loss first.

_Bardet-Biedl_

Bardet-Biedl syndrome affects many parts of the body besides the retina. It is usually diagnosed in children when symptoms of retinitis pigmentosa begin to show up during childhood. Peripheral vision loss in children who have Bardet-Biedl syndrome is followed by loss of central vision as well.

The other characteristics of Bardet-Biedl syndrome are obesity; extra fingers or toes, which sometimes are webbed; small genitals in men; irregular menstrual cycles in women; kidney disease; short stature; and mild to severe cognitive disability.

Children who have Bardet-Biedl syndrome are sometimes diagnosed with Laurence-Moon-Biedl syndrome. Laurence-Moon-Biedl is a similar syndrome which includes neurological problems but not extra digits.

_Leber's Congenital Amaurosis_

There are two syndromes to which Theodore Leber gave his name, Leber’s congenital amaurosis and Leber’s hereditary optic neuropathy, also called Leber’s optic atrophy. Leber’s congenital amaurosis is seen more frequently and is sometimes referred to simply as “Leber’s.” It is a hereditary disease of the
retina which is characterized by low vision at birth and subsequent degeneration of the photoreceptors. It is sometimes called rod-cone dystrophy because, as in retinitis pigmentosa, the rods are the first photoreceptors to be affected.

Some children who have Leber’s congenital amaurosis retain enough vision to read large print. They are usually farsighted and benefit from glasses to see objects up close. Braille is usually a preferred medium for reading and writing since vision is likely to be reduced as the children get older. Once people who have Leber’s congenital amaurosis reach young adulthood, their vision tends to stabilize.

Being hyper-sensitive to light and glare is also a characteristic of people who have Leber’s congenital amaurosis. Wearing sunshades and a cap with a dark brim is helpful, not only for comfort but to optimize people’s visual acuity.

Another hallmark of Leber’s congenital amaurosis is eye poking. Children who have this disease tend to push their fists or fingers into their eyes whenever they are resting. The result may be the deterioration of the tissues surrounding and supporting the eyes. It is difficult to change this behavior. For children who care what others think of them, subtle reminders sometimes are effective. Some children stop eye poking when they wear glasses to remind them to keep their hands out of their eyes.

In some cases, Leber’s congenital amaurosis is associated with central nervous system complications such as cognitive impairments, epilepsy, autism and motor skill impairment. Heart problems may also be part of the syndrome.

Experiments with gene therapy with people who have Leber’s congenital amaurosis are currently being conducted after success with such therapy in dogs who had this condition.

**Usher’s Syndrome**

Usher’s syndrome is a hereditary condition that causes degenerative hearing impairment as well as retinitis pigmentosa. There are three types of Usher’s syndrome, which each have different progressions in sensory loss.

The early symptoms of the first type of Usher’s syndrome are poor balance and profound deafness. Vision loss begins around the age of ten. The vision loss progresses from peripheral loss to total blindness fairly rapidly.

The second type of Usher’s syndrome begins with a moderate to severe hearing impairment at birth and typical balance. Often children who have this type of Usher’s syndrome use hearing aids and communicate through speech. They begin to lose vision as teenagers and eventually become blind. They benefit from
holding the hands of someone who is using condensed sign language when their ability to read lips is lost.

A third type of Usher’s syndrome is characterized by hearing and vision loss progressing slowly from teenage years into adulthood. Children who have it are born with typical hearing and vision and fairly good balance. By mid-adulthood they have significant hearing and visual impairments.

**Alstrom Syndrome**

The vision component of Alstrom syndrome is cone-rod dystrophy, meaning that the central vision is lost before the peripheral vision. Children who are born with Alstrom syndrome are extremely sensitive to light from early on. They often prefer to wear dark glasses even indoors. Dim light allows their rods to be active in collecting and providing visual information. Some people who have cone-rod dystrophy close their eyes after looking at a visual target because it is easier for them to see the afterimage than the actual image.

Alstrom syndrome includes other characteristics which are degenerative. Hearing loss is associated with Alstrom syndrome. It is usually mild to moderate. Children who have Alstrom syndrome are usually obese and short of stature. As adults their extra weight is usually less pronounced. Kidney and liver problems, non-insulin dependent diabetes and congestive heart failure are all associated with this syndrome. Cognitive disabilities are not part of the syndrome, but sensory losses may cause delays in social skills.

**Stargardt’s Syndrome**

Loss of central vision in childhood is the hallmark of Stargardt’s syndrome. This causes loss of color vision and loss of acuity. People who have Stargardt’s syndrome are usually identified between the ages of six and twenty years. Acuities are usually measured at their worst between 20/200 and 20/400, but a small percentage of people who have Stargardt’s have much less vision. Just as in Alstrom syndrome, photophobia may be present and sunglasses and hats with dark brims may be needed.

The gene which causes Stargardt’s syndrome has now been isolated. Gene therapy may be available in the future.

**Batten Disease**

Batten disease is a degenerative, hereditary disease which includes cone-rod dystrophy. This disease is usually diagnosed between the ages of five and eight years. The typical early signs are progressive vision loss, seizures and clumsiness. Children who are diagnosed with Batten disease have a progressive loss of cognitive ability and a personality change which creates poor social skills.
Life expectancy is shortened by this disease. Most people who have Batten disease live to their late teens or early twenties.

Research progress is being made to isolate the causes of Batten disease and trials of gene therapy and stem cell transplants are being done on animals.

Other Syndromes

There are other rare conditions that include macular degeneration or cone-rod dystrophy. If you have a student who has one of them, it will be important for you to ask the teacher of visually impaired students to provide information on that syndrome in order for you to better understand the educational implications of the condition.

References

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