



## Vision Issues for People with CHARGE Syndrome

by David Brown

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In this article I plan to provide a lengthy, but still superficial, focus on the multi-layered complexity of the vision issues faced by many people with CHARGE syndrome. There is a common tendency to assume that the actual condition of the eyes, particularly any ocular defect that might be present, is all that determines functional vision skills. While it is important to know about the condition of the eyes, of course, and to seek appropriate help from medical specialists to evaluate this and to intervene where possible, there are also many other factors that need to be understood, evaluated, and taken into consideration. As Natalie Barraga reminded us almost 40 years ago:

*'Visual functioning is related in part to the condition of the eye. More explicitly, visual functioning is determined by the experiences, motivations, needs and expectations of each individual in relation to whatever visual capacity is available to satisfy curiosity and accomplish activities for personal satisfaction.'*

For individuals with CHARGE syndrome the following would be a helpful list of things to remember when considering functional vision abilities:

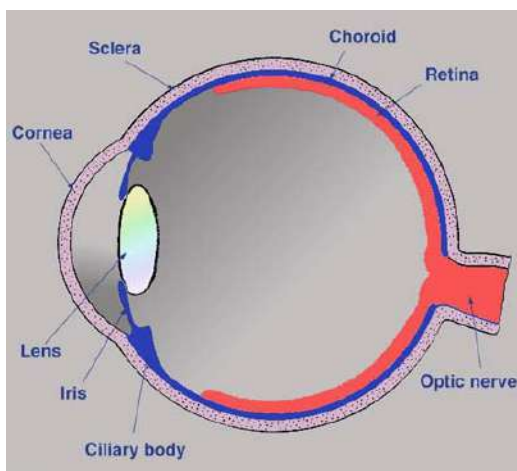
- The eyes, ocular defects, and aids to support visual functioning
- The nerve pathways that connect the eyes to the brain
- The brain itself

- Muscle tone, and the obstacles that abnormal muscle tone present to effective use of vision
- Broader issues of postural control, and energy levels, and fatigue
- Impairments in other sensory systems that will impact on visual functioning (especially vestibular issues)
- Distractibility and the place of vision in the individual's hierarchy of the senses (ie. is vision an important resource for this person, or does it seem very low priority for them?)
- Arousal levels, self-regulation abilities, and the individuals' most prevalent emotional states
- Expectations, previous experience, and motivation of the individual (and of the people around them)
- Environmental factors such as visual clutter, physical placement of things in relation to the individual, lighting levels, visual contrast, color, and other distractions that might compete for the individual's attention

As this list suggests, we need to remember that we don't see with our eyes, we see with our brains – all that the eyes can do is collect visual information for our brains to 'see' (in other words, it is the brain, not the eyes, that has to perceive, to interpret, to recognize, and then make decisions based on what the eyes are showing it). This is a big challenge for us since we all know that a brain is much more complex than a pair of eyes.

Following Barraga's comments above, we also need to remember that in any group of people with visual impairments the individual with the best functional vision skills might not be the individual with the most intact, working eyes – like so many things in life, how you use it can be as important as what you've got!

## THE EYES AND OCULAR DEFECTS



*Figure 1*

In the front-to-back cross section of the eye above (Figure 1), the front of the eye (the part that we see when we look at a person's face) is on the left side of the diagram, and the back of the eye is on the right side. Later in this article I will be paying particular attention to the retina (colored red in Figure 1), the lining which covers the inside surface at the back and sides of the eyeball. When considering ocular defects I find it useful to look at a diagram like this, then begin at the very front of the eye and work towards the back. Remember that many people with CHARGE have one eye more severely affected than the other, so what we know about one eye might not be true of the other.

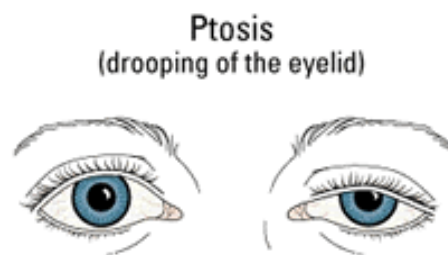
They may in effect only be seeing with one eye (monocular vision). This has implications in terms of visual field loss (ie. not seeing the full wide picture that two healthy eyes

provide), and also in terms of depth perception (difficulty seeing things like the precise width of a doorway or the exact height of a step).

### **The eyelids**

Although not part of the eye itself, over the front of the eyeball we have the eyelids. Facial palsy (cranial nerve VII) is a common CHARGE anomaly which has implications for functional vision (estimated frequency in CHARGE syndrome 40%). If the facial malformation is one-sided (unilateral) it is possible for the upper eyelid on the compressed side of the face not to open very wide and so obstruct the visual field, which is called ptosis (Figure 2), and for the lids on the other, stretched, side of the face never to close completely.

A person with ptosis will probably need to tilt their head back in order to try to see under the upper lid, possibly also using a finger to push the lid more open. Some children discover that if they watch television in the upside down position gravity pulls the upper eyelids down so that the eyes are not obstructed (Figure 3). In situations like this, even if the posture is not one that we would want the child to use for long periods of time, it is important to credit the child with doing the best they can to use their vision as effectively as possible, while we also try to work out ways in which they can be helped to use their vision more comfortably and effectively in the upright position. Sometimes, if the upper lid droops too low, surgical methods are used, either to remove part of the upper lid so that it opens wider, or to tighten the muscles that control the lid so that the same result is achieved.



*Figure 2*



*Figure 3*

If the eyelid does not close, eye drops will need to be used regularly to prevent drying out and scarring of the cornea at the front of the eye itself. Sometimes in these cases a small weight is surgically implanted in the upper lid to help gravity bring it lower and achieve full closure with the lower lid. In addition to obstructing vision, facial palsy can also result in a very inexpressive face, which adds to existing difficulties with expressive communication. A combination of bilateral facial palsy and macular coloboma on the retinas of both eyes (central vision loss) might lead to a person with no facial expression who does not appear to make eye contact, and this often leads to incorrect or lowered expectations in other people.

### **Iris Coloboma**

Coloboma is the word used to describe a part of the eye that has not formed completely. If the development of the front of the eye is disrupted during the pregnancy a coloboma

of the iris might result, and this can be seen easily just by looking at the person's eye, since the pupil is not circular as usual but seems to have an extra part to it, often referred to as a cleft, or a notch, or a 'keyhole' pupil (Figure 4).

Visual acuity and visual fields should not be affected by colobomas of the iris, but this anomaly is likely to create problems coping with brighter levels of light (photophobia), because the pupil cannot shrink in size as the level of light gets brighter. But, some people with iris coloboma show no photophobia at all, and photophobia can also sometimes be present in people with CHARGE who do not have iris colobomas in either eye. Indicators of photophobia may include



*Figure 4*

screwing up the eyes or covering them with an arm or a hand, holding the face down towards the floor all the time when outdoors in daylight, resistance to going outside in daylight, refusing to sit facing towards windows in the classroom, and refusing to face brightly illuminated computer screens. Often, the wearing of tinted spectacles or a sun visor or peaked cap can reduce the severity of many of these problems. One apparent paradox is found when people who demonstrate photophobic behavior when they need clear visual information also at other times deliberately gaze at bright light when they only need visual stimulation (which could be an indicator of their need to get all of their sensory systems reorganized due to tiredness, stress, or sensory overload). In other words, bright light can be great when it is just what the person needs, but it can be a big nuisance to them at other times.

## **Cataract**



*Figure 5*

This condition, where the lens in the front of the eye is actually an opaque milky color rather than clear and transparent (Figure 5), is rare as a congenital anomaly in CHARGE syndrome.

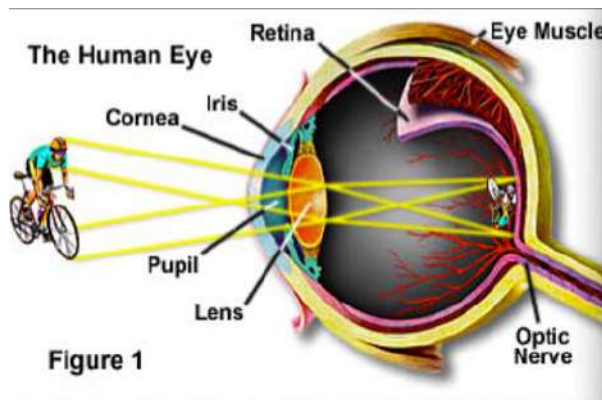
However, I have known two children with CHARGE who had very limited vision in an eye for other reasons, and had developed the behavior of staring persistently at bright light while firmly pressing a finger or a knuckle into the front or the side of the eye, presumably to

create interesting visual effects like fireworks or flashing lights. Both of these children eventually developed what were called 'traumatic' cataracts in the lens of the eye because of the heavy pressure that had been put on them over a long period of time. In one of these cases the eye was considered too fragile to try to remove the damaged lens and implant an artificial one, which would be the usual surgical procedure. In the other case the damaged lens was surgically removed, but during the operation the retina at the back of that eye detached and it was not possible to re-attach it.

## Retinal Coloboma

The retina is like a screen covering all of the back and sides of the interior of the eyeball; the retina receives visual information coming in through the front of the eye, converts it into electrical energy, and then sends this to the optic nerve which carries the information to the brain for interpretation and decision-making. If the development of the retina is interrupted during pregnancy, then a retinal coloboma will result, sometimes described as a hole or a cleft on the back interior surface of the eye where the retina should be.

Because the eyeball develops from the top downwards in a fetus, any interruption or disruption of this process is likely to leave an eye which might be intact at the top but malformed lower down, and most retinal colobomas in CHARGE syndrome seem to involve the lower part of the retina. Retinal colobomas can be very large and involve the entire retina, or they can be smaller and only involve part of the retina, there may be more than one coloboma on a retina, retinal coloboma might be present in only one eye, and when there are retinal colobomas in both eyes the size and shape and location of each coloboma is likely to be different.



*Figure 6*

Colobomas of one or both retinas will cause some visual field loss. Because the incoming image is inverted onto the retina at the back of the eye, a coloboma on the lower retina means that it is the upper part of the image that will not be seen. In the diagram below (Figure 6) the image of the cyclist is projected upside down onto the retina at the back of the eye, and the brain then reverses the image to make it match with the reality that we know about the world. But if the bottom part of the retina

is missing due to the presence of a large retinal coloboma, then it is the top part of the image of the cyclist which will be missed.

In order to see things at or above their face level (such as tall people's faces, the blackboard or the movie screen up on the wall, the book that teacher is holding up, the low hanging tree branch overhead), people with this kind of 'upper visual field loss' usually have to tilt their head back to raise their eyes just as if they were trying to look under the peak of a baseball cap which is pulled too low over their face.

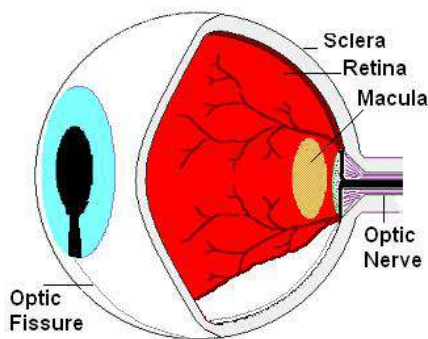


Three of the children in the photograph above (Figure 7) need to adopt this head posture when told to 'look at the camera', and we notice that the lower down the child is in relation to what they are looking at, then the more exaggerated the backward head tilt needs to be. Since the same kind of head posture is used by people with ptosis to raise their lower field of vision, and since a significant number of people with CHARGE have ptosis, then there may be two reasons why

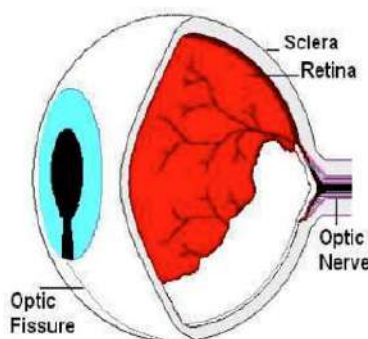


*Figure 7*

this uncomfortable and potentially unstable head posture has to be adopted. When children begin walking they may have to tilt the head back in order to see in front of them – although this posture might be crucially important for visual orientation during walking, it prevents people seeing where they are placing their feet, and it is very challenging for good sitting and standing posture and secure equilibrium. There may be extremely conflicting needs with regard to head position when walking – good balance, the need for a clear view of where the feet are being placed, and photophobia may all compel the child to flex and hold the head forward with the face down, yet the head needs to be extended and tilted back with the face up in order to really see the environment ahead.



*Figure 8.1 Upper Visual Field Loss*



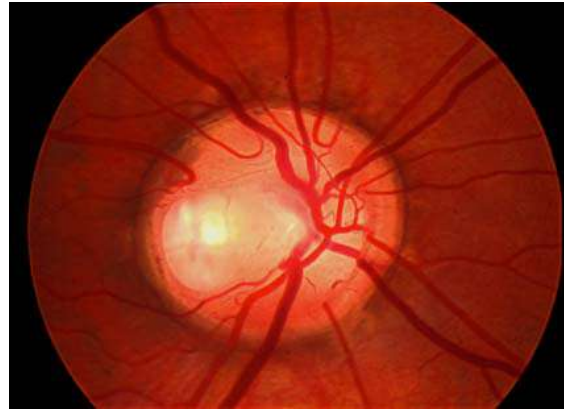
*Figure 8.2 Upper Field & Central Vision Loss*

In Figure 8.1 above, the retina at the back and sides of the eye is colored red, and a coloboma can be seen at the very bottom of the retina

where it has failed to develop during the pregnancy. The optic disc (the place where the optic nerve is attached to the retina) and the macula (the part of the retina which is used for central vision, fine visual discrimination such as reading small print or threading a needle) are not affected, so this eye will have an upper field loss but vision for seeing fine details should not be affected by the coloboma. In Figure 8.2 we can see a much bigger retinal coloboma which extends further up the eye and affects both the optic disc and the macula; this eye will have a much more extensive upper visual field loss, but, even more important, visual acuity will be affected and there may be no fine central vision at all. If the eye shown in Figure 8.2 is the only working eye that the person has then they may function as if they are completely blind apart from some awareness of light in the lower

visual field of the eye. As a result of combined field loss and very poor central vision a person may not look directly at objects or other people and may need to point their eyes to the side or above the object that they are examining to compensate by using their peripheral, or side, vision.

So the size and location of a retinal coloboma can make a huge difference to the impact that it has on visual perception and visual functioning. It is fortunate, therefore, that a retinal coloboma is fairly easy to see in an ophthalmic examination once the pupil at the front of the eye has been dilated with eye drops. Figure 9 below shows a photograph, taken through a dilated pupil, of the back of an eye which has a retinal coloboma that covers the optic disc. When this kind of examination is done the specialist can be asked to provide the patient or their family with such a photograph, or to draw a picture of what they see on the retina, so that other people can be shown in the future. It is not helpful merely to be told that a person has a coloboma if this information does not then go on to specify which eye is affected, whether it is an iris or a retinal coloboma, and the size and location of any retinal coloboma, and this information can be clarified dramatically if a photograph or a drawing is available.



*Figure 9*

### **Retinal Detachment**

Like a thin sheet of very fine cloth, the retina is held in place against the back and sides of the eyeball by pressure from the fluid that fills the eye. If the retina develops a tear or a small hole then the fluid in the eye may start to seep through the hole and the retina will gradually detach from the inside of the eyeball and float free in the fluid, which breaks the nerve connections between the retina and the back of the eye and results in complete lack of vision. If the entire retina detaches then the eye will become totally blind. Once it starts, the process of detachment can be complete within 24 hours, and surgical intervention to try to re-attach the retina is needed within a matter of a few weeks if it is to have any chance of success. Retinal colobomas carry a high risk of retinal detachment, so high impact physical activities are not recommended. Extra care is needed when imposing large rhythmic movements on a person with a retinal coloboma, or doing vigorous PE activities, and the risks of certain sports (trampoline, boxing, diving, wrestling, football, etc.) will need to be considered very carefully. Investigation of any dramatic change in a child's functioning or behavior should automatically include an ophthalmic examination in case there has been a retinal detachment.

### **Some Other Vision Issues**

Because of underdevelopment of the eyes in CHARGE Syndrome, one or both eyes may be unusually small (microphthalmia), or even missing entirely (anophthalmia). Although the presence of microphthalmia implies visual impairment, some people use their

microphthalmic eye remarkably well. People with CHARGE are likely to have refractive errors and benefit from corrective spectacles; if family members request spectacles for a person with CHARGE because they may have refractive errors it is important that they explain this clearly so that the eye specialist does not assume that the family is hoping that the spectacles will counteract the effects of retinal colobomas (which they will not). Other vision issues might include amblyopia (a weak eye), or strabismus (the eyes not working in alignment), which might be treated with surgery, or with a planned program of patching the stronger eye.

## **VESTIBULAR ISSUES**

### **The vestibulo-ocular reflex (VOR)**

We know that people with CHARGE have a very high likelihood of problems with the vestibular sense (estimated frequency 90%), and that malformations and malfunctioning of one part of the vestibular receptors in particular, the semi-circular canals, is common. In addition to monitoring all movements of the head, the semi-circular canals also organize compensatory movements of the muscles that control our eye movements, so that the fixation point of our eyes remains stable rather than moving about the same as the head; specific head movements trigger specific semi-circular canals to activate specific pairs of eye muscles in specific ways that enable this. Put very simply, if the head moves in any direction at all the semi-circular canals tell the eyes to move the same amount in the opposite direction so that they can continue looking at whatever they need to. This is a remarkably complicated but quick acting reflex sequence. The amazing thing is that although it stabilizes our visual fixation for us, we can then superimpose voluntary eye movements upon this stable base whenever we wish to. This compensatory reflex, complex and smooth and rapid, yet something that we don't need to think about at all, is called the vestibulo-ocular reflex (VOR). You can identify the reflex at work with a simple experiment. If you hold a book very still and try to read part of it as you move your head side to side and up and down and round in circles, it might not be particularly comfortable, but it is perfectly possible to keep your eyes reading and following the lines of print, thanks to your semicircular canals which are being activated by your head movements. But if you keep your head absolutely still and somebody else keeps moving the book around in front of you it is impossible to read along the lines of print since your semicircular canals are not being activated by any head movements, and your voluntary eye movements are totally incapable of keeping pace with the movements of the lines of print in the book. This gives you a small idea of what it must be like to try to use your vision when your semicircular canals are damaged, so that your eyes move away from what they are looking at every time your head moves, and then you have to search with your eyes to get them back to what they were looking at.

So there are strong links between the vestibular sense and vision. Problems with vestibular perception may affect a person's ability to maintain a stable visual field, but they may also make it difficult for the person to follow moving objects smoothly with the eyes, and to work out whether it is the object that is moving or they themselves that is moving. Not surprisingly there is often a tendency for the person with these issues to keep their head stable by getting it fully supported in any comfortable way that they can -



propping the chin on the hands and arms, or resting the head sideways on the tabletop, or getting completely horizontal on a couch or on the floor. As with any unusual postures that we observe, it is very important that we spend some time trying to work out what the behavior means to the person doing it since it might be an adaptive response that helps the person to function, rather than anything lazy or naughty.

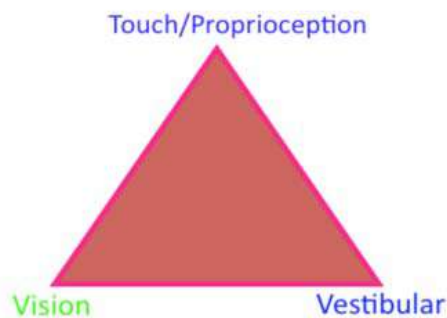
## **MUSCLES!**

When people think about muscular activities, they usually think of things like aerobics, weight-lifting, swimming, hiking, wrestling, and so on, but they rarely think about using vision, even though vision is a sense that depends upon very good control and coordination of many different muscle groups if it is to be used effectively. If we want to spend some time watching a stable object (imagine a painting in an art gallery) we need our eyes directed towards the thing we are watching and comfortably held in a stable position, we need our head stable and not in an uncomfortable unsupported position, and we need our body stable and not in an uncomfortable position. If the object we are watching is moving (imagine a tennis ball going back and forth over the net) we need an even better range of muscle control and coordination so that our eyes and head and body all move in the appropriate directions, at the correct speed, and all stop moving if the object also stops moving. On the smallest level, we need ocular muscles that can not only move and position and fix our eyes effectively but can also help the lenses in our eyes to alter their shape so that objects we look at remain clearly in focus at any distance. While all this movement is going on we also need good postural control of our bodies so that moving the eyes, and turning the head and trunk, does not cause us to lose balance and fall over. For most of us, once we are past infancy, all this happens smoothly and easily, and in an unconscious way. For many people with CHARGE, however, these are complex and challenging skills that may be impossible to master without significant adaptations to minimize the challenges.

Very persistent low muscle tone is characteristic of many people with CHARGE syndrome, and is thought to be an outcome of many possible factors including severe vestibular problems, low vision, loose connective tissue, heart defects, breathing difficulties, poor nutrition, and generally reduced sensory inputs, hence reduced perceptual awareness. The problem is then compounded by the lack of motivation to move and the resulting lack of “exercise.” Protective reactions, head control, standing, cruising, and independent walking usually develop very late for infants with CHARGE (walking happens at a mean age of between 3 and 4 years). It should not be surprising then that some children with CHARGE may appear to ‘go blind’ if their postural security is too challenged, but they may surprise us by showing some well-developed visual (and other) skills once they are flat on their back or on their side on a stable surface, with their head fully supported. This apparent paradox shouldn’t surprise us because Jean Ayres told us a long time ago that, after air to breathe, postural security (rather than using our vision!) is our next most urgent priority. Without postural security none of us is going to focus our attention on watching TV, or reading a magazine, or on carrying out a complex fine motor task like sewing or writing. First we reorganize our position to get more secure and physically comfortable, or in the most extreme situations we save ourselves from

falling, and only then do we do our TV watching, or reading, or sewing. All of our senses are designed to work simultaneously and support each other, and good equilibrium, or balance, actually depends upon interactions between three senses which form the Equilibrium Triad:

### The Equilibrium Triad



*Figure 10*

As they get older, some people with CHARGE are able to use their vision to help them to balance and stay upright because of this interdependence between these senses. They compensate for having a poor or missing vestibular sense by using the strong visual impressions made by horizontal and, especially, vertical lines in a room (for example corners, the edges of windows, doors, table tops, and wall-mounted pictures) to tell their brains which way is 'up' because the sense that normally does

this (the vestibular) is missing. So if vision is compromised or obstructed (for example when walking outside in bright light, or walking in darkness) they may have much less equilibrium because these strong visual markers are largely absent. While they are walking they may need to focus intently on using vision for balance purposes (rather like a tightrope walker staring at a stable point, almost like holding on to it with the eyes) and so not be able to transfer their visual attention to use their vision for any other activity such as watching other people walk by, or looking at the person walking next to them, or looking at the map they are holding in their hand. To do any of these latter things the person might need at the very least to stop moving and stand still, but they might also need to sit or lean against or hold on to a firm support (a tree, a bench, a lamp post, another person) to free up their vision for this other task. So it should not be surprising that some people show a reluctance to go outdoors, for example during recess at school, nor that they might have an inability to perform certain tasks when they are outdoors that they perform very well indoors. Since all fine visual skills are likely to be compromised, people who are reading might benefit from the use of a typoscope (a letter-box shaped frame) which will isolate one single line of text at a time. Similarly, the use of large print on a computer might be very helpful to a person, not necessarily because their visual acuity is poor but because they need help to isolate the line of text on which they should be visually fixating.

### VISION AS A COMPENSATORY SENSE

We know that many people with CHARGE have difficulties with self-regulation, often described as extreme mood swings for no apparent reason. We also know that there is a high incidence of anxiety in the population of older children and adults with CHARGE, which can create significant difficulties in coping with transitions, and can also produce ritualistic behaviors that look similar to Obsessive Compulsive Disorder. Although all

these features of CHARGE syndrome are considered to be largely neurological in origin, we need to remember the huge challenges that are posed by multi-sensory impairment and the fact that most people with CHARGE are trying to function with fragmented, variable, and missing sensory information. Like any other damaged sense, impaired vision can be exhausting to use, especially for long periods of time, and particularly when it is combined with many other health and sensory issues. Yet, in spite of this, it is noticeable that many people with CHARGE work very hard to use their residual vision since it can fulfil so many vital functions for them – in seeking re-assurance from the world around them (everything exactly where it is supposed to be), in anticipating future events (the magic of the picture calendar and the schedule board), in ‘reading’ the behavior of other people, in modulating arousal levels and self-regulating for improved functioning (the magic of sensory breaks and the spinning ceiling fan!), and in locating themselves securely in space and in the physical environment because their vestibular and proprioceptive and touch senses are not able to do this very effectively.

### **Other resources**

The CHARGE Syndrome Foundation provides a great deal of useful information free on its website ([www.chargesyndrome.org](http://www.chargesyndrome.org)). Particularly recommended are the CHARGE Syndrome Professional Packet, the CHARGE Syndrome Manual, the American Journal of Medical Genetics articles featured in the issue dedicated to CHARGE Syndrome, and the ASHA CHARGE Syndrome web program. There are also two interesting webcast videos (one an overview of CHARGE, and the other a consideration of the impact of CHARGE on communication needs and learning styles) both of which can be accessed free at <http://www.perkins.org/resources/webcasts/> . See also the article on the vestibular sense reprinted from Deafblind International Review on this website.

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