CVI and CHARGE

by David Brown
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Recently several people have asked me if Cortical Visual Impairment (CVI) is part of the spectrum of anomalies in CHARGE syndrome, or if it is possible to have both CHARGE syndrome and CVI. In an article about vision issues in CHARGE syndrome that I wrote for our CDBS newsletter reSources (Winter 2010), I originally included a paragraph about the possible presence of CVI in CHARGE, but the article grew too large and in the end I had to edit it out. Today another question about CHARGE and CVI cropped up, so I thought I would finally return to the issue and put down some of my thoughts.

First, there is obviously no reason to say that if you have CHARGE syndrome then you cannot also have CVI (or autism, or mental retardation, or dyslexia, or bulimia, or arachnophobia, or blonde hair…etc.) When we consider the brain malformations and other neurological and sensory processing issues in CHARGE then it actually seems quite likely that some of these children might have some degree of CVI, in addition to their other visual disabilities.

I think the question is made especially complicated, though, by the very common vestibular issues that we see in the syndrome, plus the fact that ALL sensory channels are likely to be damaged, missing, or compromised. With a poor or absent vestibular sense, it is going to be very difficult to fixate visually on a target and maintain fixation without first getting the body and the head firmly supported (e.g., on the tabletop or floor, or against a doorpost). I think sometimes a child in this situation might have to look and visually locate a target first, then look away so that they can concentrate on moving towards it or reaching for it without also having to continue working hard to keep their body, head and eyes stable for looking. This is identical to one of the classic CVI behavior patterns, but for very different reasons (CVI-type behavior #1).

The same issues with the vestibular sense can make it hard to scan the environment visually in a smooth and well-organized way to search for and locate a desired object; hence I think that children with CHARGE sometimes move in closer to eliminate the more complex wider 'picture', so that visual scanning requires less movement of the head and eyes (CVI-type behavior #2).

For these reasons, plus others we do not understand yet, many people with CHARGE need an unusually long processing time for perceiving through various sensory channels, so there might be a very noticeable latency or time delay in perceiving and then responding to many kinds of sensory inputs (CVI-type behavior #3). Eye contact and facial regard is often delayed or missing (CVI-type behavior #4) - maybe because the human face is so complex in multi-sensory terms - but also possibly because of other emotional or psychological issues not specifically related to

functional vision impairment. Because of the visual fixation and tracking and scanning problems that result from vestibular dysfunction, another person's head and face is likely to be an impossibly challenging object to regard for any length of time unless it is absolutely stable, and none of us holds our head and face that way. I like to test for this by seeing how/if a child will look at my face while I am upright and then seeing if they will do it when my head is completely stable by resting it on a tabletop or floor, or cupped in my hands.

Basically, I am saying two things: 1) Yes you can have both CHARGE and CVI, but I don't believe that this is a very common combination, and 2) The multi-sensory issues in CHARGE, especially the very prevalent vestibular issues, are almost certainly resulting in a variety of functional adapted visual behaviors that parallel what we see in many children with CVI, but that have a different origin.

As is so often the case lately, I find myself wading into uncharted waters. If anybody has ideas, anecdotes, or references on this issue, I would love to hear from them.