Cortical visual impairment

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Cortical visual impairment (CVI) is emerging as one of the important causes of blindness or visual impairment in India. This increasing trend is likely to continue as further advances in the field of neonatology and fetal medicine increase the chances of survival of a premature infant or an infant with multiple congenital abnormalities. Previous studies from developed nations have shown that infants born prematurely and those with low birth weight are at a risk of severe ophthalmic impairment.

What is CVI?

The current definition of cerebral or CVI includes all visual dysfunctions caused by damage to or malfunctioning of the retrochiasmatic visual pathways in the absence of damage to the anterior visual pathways, or any major ocular disease.

We need to remember that CVI does not imply total blindness and is a spectrum of vision loss ranging from mild visual impairment to complete visual deprivation.

Pathogenesis

The central nervous system in a newborn is susceptible to various damaging influences such as hypoxia–anoxia, hypoglycemia or a combination of both. Two distinct subgroups of cortical visual loss in terms of the areas of damage and the time of injury have been described. Injury in full term neonates, predominantly involves the striate and peristriate cortex, whereas in preterm neonates, injury involves the subcortical white matter, including the optic radiations. This results in different patterns of damage as seen on neuroimaging and also results in different clinical manifestations.

In a full term neonate, the vascular supply of the brain is derived from the major cerebral arteries and its watershed areas lie at the interfaces between the major cerebral arterial distributions. Hence, any hypoxic-ischemic injury produces watershed infarctions in the parieto-occipital and parasagittal cortex, resulting in cortical visual loss. Whereas, in the developing brain, the cortex and underlying white matter receive their blood supply from branches of the blood vessels on the surface of the cerebral hemispheres and the watershed zone lies within the periventricular white matter.

Hence, preterm injury to the brain results in injury to the subcortical white matter, resulting in periventricular leukomalacia (PVL). Other causes of injury in neonates include birth trauma, intraventricular hemorrhage and hypoglycemia. Neonatal hypoglycemia, in particular, is known to cause significant visual deficits and neuroimaging in these cases shows restriction on diffusion predominantly in the occipital areas.

Causes

A wide variety of insults to the central nervous system can result in CVI. These include: perinatal conditions such as birth trauma, hypoxia–anoxia, hypoglycemia and post-natal conditions such as sepsis, meningitis, encephalitis, accidental and non-accidental trauma, cerebral malformations such as Chiari malformations, Dandy–Walker complex, hydranencephaly, porencephalic cysts, metabolic and neurodegenerative conditions such as MELAS (mitochondrial encephalopathy, lactic acidosis and stroke-like episodes), Fabry’s disease, Leigh’s disease, X-linked adrenoleukodystrophy and hydrocephalus.

Clinical presentation and diagnosis

As mentioned previously, CVI can manifest as mild visual loss with a slight delay in achievement of visual developmental milestones or as near total or total blindness. These babies are most frequently brought to the ophthalmologist with complaints of “not seeing the mother’s face”, poor visual response to light, “shaking of eyes” or nystagmus.

These children often have multiple other disabilities and one must remember to make the child comfortable before starting the examination. Responses to bright light, colored objects and human faces must be assessed. Some of these babies may respond better in a supine position and dim light.

One must look for the presence of nystagmus, strabismus and ocular motility deficits and pupillary responses. Cycloplegic refraction must be done along with a thorough fundus examination.

Nystagmus is rare or absent in children with CVI. However, roving eye movements or occasional bursts of nystagmus may be seen and the presence of such eye movements signifies severe visual impairment. An intact geniculostriate pathway is a prerequisite for the development of congenital nystagmus and hence nystagmus is absent in extensive posterior pathway disease. However, some children with combined anterior and posterior visual pathway disease may present with CVI and nystagmus.

Strabismus is common in children with CVI. Both esotropia and exotropia have been reported. Esotropia is more common in children with PVL and may be associated with latent nystagmus, dissociated vertical deviation and ‘A’ pattern. Other studies have shown that cortical visual loss is associated with exotropia more commonly than esotropia. Also, children with more extensive
neurological damage may have congenital exotropia.10,12

Other ocular motor abnormalities that have been noted in these children include tonic downgaze, horizontal conjugate gaze deviation and defective smooth pursuits and saccadic eye movements.10,13

Fundus examination is extremely important to detect the presence of associated optic atrophy and also to rule out other conditions with similar clinical presentation such as retinal dystrophies, congenital macular scars, sequelae of retinopathy of prematurity, etc.

In cases where the pathological processes affecting the posterior visual pathways and the cerebral cortex have not affected the anterior visual pathway, the fundus is usually normal and the disk appears healthy. However, studies involving children with retrogeniculate vision loss, have described a variety of optic nerve abnormalities including optic atrophy, optic nerve hypoplasia and pseudoglaucomatous cupping.4,12

In additional to structural damage, one must keep in mind that these children also have signifiant functional deficits such as visual field defects, which include generalized field restriction, central scotomas, homonymous hemianopias and altitudinal defects.10,12

One must keep in mind that vision is a complex function and has multiple components. These children may show other visual functional abnormalities which may be difficult to quantify, such as difficulties in visual processing, difficulty in simultaneous perception and “crowding” phenomenon. Other peculiar visual traits include a tendency to stare at bright objects, such as fluorescent room lights or the sun. This is known as “light gazing” and is regarded as a sign of severe visual impairment.14

These children also, often function better in a familiar environment, may show a better visual performance for objects in motion as compared with static objects and are better at identification of colors as compared to their perception of form.15

Some children also complain of photophobia.

Diagnosis is confirmed by neuroimaging. While imaging of the infant brain is possible by ultrasound and computed tomography, magnetic resonance imaging (MRI) is the preferred modality. MRI can help to pick up even the subtle changes of PVL such as abnormal dilatation and irregularity of the lateral ventricles, high intensity signals in the periventricular white matter on T2 weighted images and periventricular gliosis. Severe hypoxic injury may lead to encephalomalacia with surrounding gliosis.10 Other abnormalities seen include thinning of the corpus callosum, altered signals from the thalamus and putamen and cerebellar atrophy.10,12

It is not clearly known to what extent PVL and CVI and the associated neurological abnormalities affect visual evoked potentials (VEP), but in general, the absence of a flash VEP is associated with a poor prognosis.10

Electroretinogram may help by ruling out retinal dystrophies, which may sometimes present a similar clinical picture.

Management

Early diagnosis and intervention is the key to management in these cases. A team approach integrating the services of a pediatric ophthalmologist, pediatrician, physiotherapist, speech therapist and vision therapist is desirable. Parental counseling is of utmost importance and one must remember that while it is definitely important to provide the parents a realistic picture of their child’s condition, encouraging the parents to start rehabilitation and remain positive in their outlook may go a long way toward bettering the child’s life.

References