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Diagnosis and Care of Children With Cerebral/Cortical Visual Impairment: Clinical Report

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abstract

Cerebral/cortical visual impairment (CVI) is a leading cause of pediatric visual impairment in nations with developed economies and is increasing in those with developing economies. Because vision is the predominant sense used for learning, delay in diagnosis of CVI can negatively affect education, making early detection and management important. The American Academy of Pediatrics has published the policy statement "Visual System Assessment in Infants, Children, and Young Adults by Pediatricians" and an accompanying clinical report that are based on identifying potential causes of ocular visual impairment in children. Yet, routine vision screening may not accurately identify the brain-based visual impairment in children with CVI. Moreover, children with CVI often have medical complexity with other neurocognitive impairments and serious medical conditions that can make the diagnosis of CVI more difficult. Strategies are necessary for early identification of CVI to promote early diagnosis and referral for vision services that may allow a child with CVI to engage more fully in school, activities of daily living, vocational pursuits, and recreational activities. Knowledge of the characteristics of CVI as well as risk factors for CVI will assist the pediatrician in identifying children with CVI. This clinical report is complementary to previous vision screening policies, allowing both ocular and brain-based visual impairments in children to be identified and addressed. Pediatricians, other primary care physicians, pediatric ophthalmologists, neurologists, and other specialized pediatric eye care clinicians can identify children with CVI and coordinate effective evaluation, diagnosis, and referrals for vision services for these children.

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BACKGROUND

Cerebral, or cortical, visual impairment (CVI) is a disorder characterized by visual deficits resulting from damage to visual pathways in the brain (rather than the eye) that are posterior to the lateral geniculate body, including visual processing areas. Because of improved survival of children with neurologic deficits and advances in the management of ocular blinding diseases, CVI is a leading cause of pediatric visual impairment in the United States and other nations with developed economies.[1] CVI is also emerging as a significant threat to vision in regions of the world with developing economies. In 2019, a study from a tertiary eye care center in South India reported that 44% of children younger than 3 years with profound visual impairment were diagnosed with CVI.[2] Furthermore, the prevalence of CVI may be underestimated, especially in children with special needs or disabilities. A recent study in the United Kingdom found that 31.5% of children who required extra educational help or screened positive on CVI questionnaires exhibited characteristics of CVI.[3] Another study from the United Kingdom demonstrated a high incidence of CVI in children of any ethnicity historically regarded as minorities.[4] These data raise concern for equity in diagnosis and services for children in minoritized populations with CVI. The early identification, treatment, and prevention of disability from CVI often requires a multidisciplinary approach involving pediatricians, pediatric neurologists, pediatric ophthalmologists, developmental–behavioral pediatricians, neurodevelopmental pediatricians, and other pediatric health care clinicians.

DEFINITION

Many experts use the definition for CVI proposed by Sakki et al[1]: "a verifiable visual dysfunction which cannot be attributed to disorders of the anterior visual pathways or any potentially co-occurring ocular impairment." This definition emphasizes a critical feature of CVI: Either there is no ocular abnormality, or the child's visual function appears worse than expected based on the degree of ocular pathology. However, there is debate in the literature regarding what constitutes "visual dysfunction." The conservative definition of CVI, which is consistent with the World Health Organization's definition of visual impairment,[2] requires reduced visual acuity or a visual field defect (in patients who can participate with accurate testing) for diagnosis. However, some practitioners diagnose CVI in patients with abnormalities of higher-order visual processing (such as deficiencies in recognizing objects or processing motion) and who still retain normal visual acuity and visual fields.[3] Recently, the National Institutes of Health (NIH) CVI Working Group defined CVI as "a spectrum of visual impairments caused by an underlying brain abnormality that affects the development of visual processing pathways and is characterized by deficits in visual function and functional vision." The group further noted that "the visual dysfunction in CVI is greater than expected by any comorbid ocular conditions ... [and] may manifest as lower or higher-order afferent visual deficits, or both, leading to characteristic behaviors in affected individuals."[5]

Cerebral visual impairment and cortical visual impairment are often used interchangeably. There is also debate about these terms. Cerebral is used more frequently in Europe, and cortical is more frequently used in the United States. Some experts describe cortical visual impairment as a distinct entity with specific characteristics.[6] In this report, the term cerebral/cortical visual impairment is used as a general term to be consistent with the terminology used in the recent NIH working definition of CVI.[5]

ETIOLOGY AND COMORBIDITIES

CVI may develop in any child with a neurologic disorder affecting the higher-order visual pathways in the brain. The most frequent etiology of CVI is hypoxic-ischemic encephalopathy.[7,8] Other common presumptive etiologies of CVI are listed in Table 1.[9]

Because of the proximity of the optic radiations to the corticospinal tracts, cerebral palsy is a frequent comorbidity, especially in children who were born preterm. The reported rate of CVI in children with cerebral palsy ranges from 26% to 83%, depending on the definition of CVI.[10] Authors report that 21% to 47% of children born preterm have characteristics consistent with CVI.[11] Other comorbidities in children with CVI include intellectual disability, developmental delays, sensorineural hearing loss, microcephaly, and neurodevelopmental disorders such as autism spectrum disorder.[7,12] The relationship between CVI and autism spectrum disorder is complex, however, because certain visual deficits (such as prosopagnosia: inability to recognize faces of familiar people) in children with CVI may lead to abnormal social interactions that simulate symptomatology of autism spectrum disorder.[13] Diagnosis of autism spectrum disorder in children with visual impairment may require a specialized approach.[14]

NATURAL COURSE AND PROGNOSIS

Children with CVI are not a homogeneous group with respect to their underlying cause and degree of physical and visual disability. Assessing vision in children with multiple disabilities can be difficult, and their vision responses in a clinical setting may not be a true representation of their function in more familiar environments. Parameters such as standard neuroimaging (structural MRI) and electrophysiologic testing (visual evoked potentials, electroretinography) have not proven to be consistent diagnostic and predictive tools.[9] Functional MRI and tractography (diffusion tensor imaging) may be promising for the future.[15]

TABLE 1 Common Potential Etiologies of Pediatric CVI
Central nervous system malformations (eg, schizencephaly)
Head trauma
Hydrocephalus
Hypoxic-ischemic encephalopathy
Infections (eg, meningoencephalitis)
Metabolic and genetic disorders (eg, hypoglycemia, leukodystrophies)
Seizures (especially infantile spasms)
Toxins and drugs (especially in utero drug exposure)
Intraventricular hemorrhage and periventricular leukomalacia in prematurity

No specific underlying cause of neurologic dysfunction has been found to be predictive of outcome.[1] Where they start and how they respond to therapy is often unique to the individual, but several studies show that most children with CVI will have some degree of improvement in visual function during childhood.[7,16]

CVI AND EDUCATION

Vision is an important component in the development of sensory, language, motor, cognitive, and social skills. Exposure to visual experiences forms mental representations that allow children to learn through imitation and interaction during the most formative years of early childhood. Visual experiences are needed to develop visual acuity, peripheral vision, contrast sensitivity, and perception of faces.[17] Early impairment to the cortical vision centers may have great neurodevelopmental consequences given their importance on multiple developmental domains and learning. Studies in typically developing children consistently report associations between visual impairment, learning challenges, difficulty with social relationships, and lower quality of life. In later childhood, CVI may impede the ability to fully attain preacademic or academic skills, because visual processing challenges may impact learning. Amplifying and fully understanding the learning challenges associated with CVI is complicated by the impact of cooccurring conditions such as epilepsy, intellectual disability, and cerebral palsy. Children with CVI may require high levels of tailored support to address cognitive, communication, and social and independent living skills.[24] Earlier identification and intervention may positively affect school success. Early intervention (EI), vision services, early education, and school-based interventions may contribute to later success.

IDENTIFICATION OF CHILDREN WITH CVI IN PRIMARY CARE ENVIRONMENT

History

Parent/caregiver interviews and histories are important in the evaluation of a child with possible CVI. Medical history taking can identify conditions presumptively associated with CVI such as preterm birth, hypoxic-ischemic encephalopathy, epilepsy, etc (Table 1), raising clinical suspicion. Good history taking can reveal a pattern of behaviors consistent with CVI, but these behaviors can vary depending on the level of visual impairment. Children with CVI can have profound visual impairment or have near-normal functional vision.

It is important to query families about the child's vision and their concerns. Behavioral characteristics may also raise concern for CVI (Table 2), although some may not be specific to this diagnosis.[8] Children with CVI may experience significant day-to-day variability in visual function and require increased time to respond to a visual stimulus (latency). They may exhibit light gazing (preference for looking at lights) or aversion to light. Motion may be necessary for visual response in some patients (eg, the child does not see a toy until the caregiver shakes it). Children with CVI may look eccentrically while reaching for objects. They may also have more difficulty identifying objects in a complex visual environment, which may manifest as inability to find a specific toy in the toy box or an item of clothing in a pile of clothes.[25] Finally, recognition of specific objects or familiar faces may be impaired.

Physical Examination

Pediatricians can refer to the recommendations in the American Academy of Pediatrics policy statement "Visual System Assessment in Infants, Children, and Young Adults

TABLE 2 Characteristics of CVI
• Challenge or apparent disinterest in locating and sustaining visual focus on objects
• Need for extra time to respond to visual stimulus (latency)
• Affinity for light in some children while others display light sensitivity
• Movement of a visual object (extra sensory cue) may enhance detection of the object by the child although gross motion perception may be reduced
• Unusual visual behaviors (eg, not looking at object when reaching for the object) (eccentric viewing)
• Challenges with interpreting complex visual objects, arrays, or surroundings
• Challenge with recognition of objects or faces
• Preference for bright colors
• Variable visual response based on environment, fatigue, distraction, etc

These characteristics may aid in identifying children at risk for CVI. The characteristics may change and improve over time.

by Pediatricians"[26] when assessing the vision behaviors of their patient. Observation of characteristic behaviors listed in Table 2 may increase the index of suspicion for CVI. Pediatricians should be aware that children suspected of having CVI may also have comorbidities of ocular visual impairment that also warrant further evaluation and treatment by a pediatric ophthalmologist. Children at risk for CVI because of history, behavioral characteristics of CVI noted on physical examination, or abnormalities on the aforementioned visual assessment should be referred for pediatric ophthalmologic evaluation.

Ocular Examination

When a child suspected of having CVI is identified, a referral to a pediatric ophthalmologist enables a more in-depth assessment to assist in establishing the diagnosis of CVI. A pediatrician with concerns about a child's visual function may request an evaluation for vision services, especially if a delay in seeing a pediatric ophthalmologist is expected because of access issues.

An ophthalmologic examination will also seek to identify and treat additional ocular causes of vision loss that may exist as a comorbidity. Because there is no standard protocol for diagnosis, ophthalmologists generally diagnose CVI when visual function is worse than expected based on the degree of ocular pathology. If there is uncertainty about the diagnosis, additional consultations with a pediatric ophthalmologist with specific experience with CVI, a pediatric neuro-ophthalmologist, and/or a specialist in neurodevelopmental disabilities may be helpful. An MRI or computed tomography scan of the brain may be obtained, as well as further evaluation by a pediatric neurologist.

When a diagnosis of CVI is made, the pediatric ophthalmologist can refer a child for an evaluation for vision services from the appropriate state service, if not already done by the pediatrician or other provider. CVI interferes with a child's ability to access his or her educational materials providing medical necessity for vision services regardless of degree of disability.

VISION SERVICES

In the United States, vision services are distributed by each individual state. Each state has its own definition of eligibility based on acuity, visual field, or the diagnosis of a chronic condition causing disability.[27] Educational resources through the state or nonprofit organizations support families by providing equipment and educational planning. A teacher of the visually impaired (TVI), an educator with special certification in educating those with visual impairment, administers these services.

A TVI can assess a child's visual function and make recommendations for the classroom that address the specific characteristics of CVI demonstrated by the child and work to provide accommodations. Services may be accessed through EI programs for young children or through the local school district for children 3 years or older. TVIs prepare functional vision assessments, learning media assessments, and assistive technology assessments.[28] The functional visual assessment identifies a child's ability to use vision to complete tasks. A learning media assessment determines how children use their senses to access educational materials. Assistive technology assessments determine devices to support a child's needs. This information may allow better access to educational materials.

SERVICES

Children with CVI have a range of challenges that require long-term intervention and monitoring. The medical home can be a starting point for advocacy to ensure children with CVI have access to routine medical and ophthalmologic care, needed therapies, and participation in programs that support developmental monitoring, EI, and special education.[29,30] Children with CVI may be eligible for services from agencies located in the community, from the state, or from national organizations. Unfortunately, the availability of services can vary at the local, county, and state levels. Thus, it is important for the pediatrician to help families seek out the appropriate services available in their specific region. An important consideration when advocating for children with CVI is the inclusion of a TVI in intervention planning. After a request for vision services has been made through EI, the state, or school district, a TVI will visit the patient and perform an evaluation.

Specialized multidisciplinary care programs, such as high-risk infant follow-up programs, often located in academic centers, can provide medical and developmental assessments, ongoing monitoring, parent/caregiver support, hearing assessment, ophthalmologic assessment, care coordination, and home assessment for infants born preterm (<32 weeks), very low birth weight infants, or infants with a range of risk factors. Services typically begin after discharge from a NICU and can be provided to 3 years of age.

EI is a federal grant program for infants and toddlers with a disability or who are at risk for developmental delay or disabilities. It is funded through Part C of the Individuals with Disabilities Education Act.[31] Part C assists all states and territories in providing critical intervention to children younger than 3 years for the purpose of enhancing future learning. Depending on the state, these programs are offered through state departments of education, health, human services, or rehabilitation or through independent agencies contracted by a state system. EI programs are voluntary. No-cost services offered for children include screening, evaluation, and determination of eligibility by mandate occurring within 45 days of referral. Children with CVI are often eligible for EI and may receive developmental enrichment, coordination, health, and social services, as well as services when they reach 3 years of age and progress to preschool services. An individualized family service plan must be drafted to delineate the services that the child and family will receive. Services are often provided to the family in their home or at a local community setting.[31] Pediatricians are encouraged to be familiar with their state's policies regarding the definition of developmental delay and the agencies in their community providing EI services and to develop working relationships with EI providers to best advocate for their patients and families.

Special education services are provided through Part B of the Individuals with Disabilities Education Act and are available to all students 3 to 22 years of age, with the right to a "free, appropriate, public education," regardless of disability. Moreover, these services must be provided in the "least restrictive environment" appropriate for the child.[31] Also included is the right to be in the same classroom as children without disabilities. Under Part B, families must be included in the process of developing a plan to address their child's individual needs. The individualized education program is developed by the family with representatives from the school and special education department. This contract sets the learning goals, monitoring, and support needed to achieve the goals set forth by families. Public schools are also responsible for providing accommodations or adaptations in the classroom to support students with special vision needs.[31] Families may be encouraged to learn about the full complement of services including vocational skills training that a school district might provide, as well as alternative educational settings that may better serve their child with CVI.

NEEDS OF OLDER CHILDREN AND YOUNG ADULTS

Evidence demonstrates that transition of care from the pediatric to the adult health care system for children with developmental delay maintains important accommodations and interventions that may contribute to optimal outcomes.[32] Yet, there is little evidence-based research concerning transitional services for patients with visual impairment. Familiarity with CVI among adult health care clinicians may be lacking. Accommodations ensure educational opportunities continue with the transition to higher education and in adult life. Vocational training, independent living skills training, and adaptive aides are available to families through federal- and state-funded programs and centers.[33] Timely planning is necessary for approval for accommodations for standardized testing or an individual plan for employment and attention given to ensure continued resources for educational, vocational, and recreational pursuits. Assessment for readiness for using public transportation and driving serves to promote increased independence for some children and young adults. Establishing needs for individuals with CVI and partnering early with adult care institutions and organizations can help older children and young adults continue access to needed services. The TVI may assist in preparing a list of needs on the basis of current accommodations.

FUTURE DIRECTIONS IN DIAGNOSIS AND TREATMENT OF CVI

A pediatric ophthalmologist plays an important role in the assessment of a child with suspected delays in vision development. Visual function in children with developmental delays and in nonverbal children is often very challenging to assess. Moreover, standard assessments of visual acuity may fail to capture the myriad other aspects of visual function that are affected in CVI. Thus, more accurate measures of visual acuity and visual function in children with CVI are under investigation. These include preferential looking tests, sweep visual evoked potentials, functional vision assessments, neuropsychological tests of visual perception, and eye tracking strategies.[34] Further research is necessary to validate an objective, quantitative, comprehensive, and scalable method of visual function assessment in children with CVI for use in both clinical and research settings.

There is currently no evidence-based treatment targeted specifically for CVI.[9] EI is desirable because of age-dependent neuroplasticity,[35] but the optimal timing and type of intervention has not been proven. Some measures of visual function (functional vision assessment, formal eye tracking technology) may eventually provide direction for individualized treatment strategies. Longitudinal controlled studies are required to assess the effectiveness of proposed treatments. In the meantime, the assessment and management of children with CVI require a multidisciplinary approach to facilitate access to appropriate services and accommodations throughout childhood and beyond.

RECOMMENDATIONS CONCERNING PEDIATRIC CEREBRAL/CORTICAL VISUAL IMPAIRMENT

- Early identification of children with CVI: Knowledge of presumptive causes of CVI and directed history taking about visual behavior can help pediatricians and other clinicians identify children with CVI and refer them for ophthalmologic and neurologic examination, ideally a clinician with expertise in children who will establish the diagnosis and refer for further evaluation for vision services by a TVI and other appropriate services. More evidence-based research is needed in validating screening tools to detect early signs of CVI.
- Improve communication among child's health care team: Sharing of information between the family and the rest of the child's team, including the TVI, may allow a fuller understanding of the child's situation by all parties. Communication and identification of other medical or neurologic conditions, disabilities, and possible other causes of ocular visual impairment allows for better multidisciplinary care and treatment planning.

- Address disparity in medical care: There is a higher risk for children from minoritized groups to have conditions predisposing to developmental delay and visual impairment. Equitable access to care and universal screening may address ongoing disparities.
- Optimize transition of care planning for young adults with CVI: Early planning and identification of special vision needs allows for better access to services when children with CVI reach adulthood.
- Support research in CVI: Funding for evidence-based research concerning CVI is needed to validate screening, diagnosis, and treatment modalities. A validated quantitative measure of CVI severity may aid in planning of vision services and monitoring of progress. Evidence-based research may stimulate additional financial support for services for those with CVI.
- Improve educational resources regarding CVI: Pediatric ophthalmologists, pediatric neurologists, TVIs, occupational therapists, developmental–behavioral pediatricians, neurodevelopmental pediatricians, and all members of the multidisciplinary team that provide services to children with visual impairment can learn more to identify and support children with CVI. Consideration of increased concentration on CVI in curricula may be one strategy to support this goal. CVI is one of the most common causes of pediatric visual impairment in developed economies. Improved public health policies and education are important to optimally support children with CVI, their families, and those who provide care for them.

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ABBREVIATIONS

CVI: cerebral/cortical visual impairment
EI: early intervention
TVI: teacher of the visually impaired

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