CHARACTERISTICS OF VISUAL FUNCTIONING IN INDIVIDUALS WITH PROPIONIC ACIDEMIA: A CASE STUDY

VALENTINA MARTAĆ*, VESNA VUČINIĆ, BRANKA JABLAN

University of Belgrade, Faculty of Special Education and Rehabilitation, Visokog Stevana 2, Belgrade, Serbia,
contact: valentinamartac@fasper.bg.ac.rs

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Abstract: *Propionic acidemia is a rare autosomal recessive metabolic disorder caused by a deficiency of the enzyme propionyl-CoA carboxylase, leading to the accumulation of toxic levels of propionic acid in the blood. This accumulation can result in life-threatening metabolic crises and a wide range of systemic and neurological complications. In individuals with propionic acidemia, recurrent metabolic decompensations, altered neurological status, developmental delays, and psychomotor retardation are commonly observed, with significant variability in their clinical presentation. Visual impairment in propionic acidemia may arise due to the impact of these associated systemic and neurological disturbances on the optic nerve, visual pathways, and cortical visual processing areas, potentially leading to cerebral visual impairment. This case study presents the visual functioning of a seven-year-old girl with a confirmed diagnosis of propionic acidemia. It highlights her specific visual profile and describes individually designed adaptations that support visual access and processing. Observations over time in different settings revealed patterns such as delayed visual responses, fluctuations in visual performance depending on the time of day, color preferences, difficulties in object recognition within visually cluttered scenes, and stronger responses to moving versus static visual stimuli. Based on these findings, a targeted visual stimulation and (re)habilitation program was created, along with an individualised adaptation plan to enhance visual learning and interaction with the environment.*

Keywords: *visual impairment, metabolic disorders, cerebral visual impairment, visual behavior, case study*

INTRODUCTION

Propionic acidemia (PA) is a rare, inherited disorder of protein metabolism that typically manifests in early infancy with the onset of ketoacidosis and encephalopathy; this disease consistently leads to various chronic complications (Alvarez et al., 2015; Arias et al., 2014). The first case of PA was described in 1968 (Hommes et al., 1968). The global incidence of this rare disease ranges from 1:50,000 to 1:100,000 (Al-Hamed et al., 2019). PA is particularly prevalent in specific populations, such as the Inuit people living in Greenland, where the incidence is as high as 1:1,000 (Ravn et al., 2000). Although there is currently no cure for PA, timely and accurate diagnosis, along with appropriate treatment, can stabilise the disease and prevent severe complications (Zhang et al., 2023).

The clinical spectrum of PA is broad, ranging from early manifestation during the neonatal period to later onset PA in infancy or early childhood. Neonatal PA is the most common form of this disease, and it is characterised by the fact that an apparently healthy newborn develops poor feeding and reduced alertness within the first few days of life, followed by progressive encephalopathy of unclear origin. If the disease is not diagnosed promptly - often through newborn screening - and appropriate treatment is not initiated, the condition may progress to encephalopathy, manifesting as lethargy, seizures, or coma, and can result in death (Aima et al., 2012). The deficiency of the enzyme propionyl-CoA carboxylase (PCC) leads to the accumulation of intermediate products - methylmalonate and propionate - as well as other toxic derivatives. In addition to early-life encephalopathy and ketoacidosis, this accumulation can also cause chronic vomiting and psychomotor delay (Alvarez et al., 2015; Williams et al., 2009). The encephalopathy itself leaves lasting effects

on overall development, and the brain lesions observed in diagnostic imaging often resemble those caused by stroke ("stroke-like"; Alvarez et al., 2015). Acute metabolic decompensation, marked by the buildup of toxic metabolites, presents as a "metabolic crisis" that results in the deterioration of the general condition (Alvarez et al., 2015; Williams et al., 2009). While the clinical symptoms and diagnosis of PA are well established, the underlying pathophysiological mechanisms remain poorly understood. This may be one of the reasons why, aside from dietary restrictions, there is currently no robust set of effective therapies for PA (Marchuk et al., 2023). The clinical manifestations of PA, which may appear during the neonatal period or later in development, can include growth retardation, cognitive dysfunction, epileptic seizures, basal ganglia lesions, pancreatitis, cardiomyopathy, and chronic kidney disease. Other reported complications include optic atrophy and sensorineural hearing loss (Aima et al., 2012). In a sample of 55 patients with PA, ranging in age from 5 days to 18.6 years, who were treated at 16 metabolic centres in Germany, Austria, and Switzerland, the majority exhibited mild impairment in psychomotor and cognitive development. Among the preschool-aged children (≥ 3 years, n = 45), 44% attended specialised kindergartens for children with developmental disabilities. In the school-aged group (≥ 6 years), 70% (21 out of 30) required special education services. Only five children attended regular schools, and among them, only one was enrolled in a school that offers qualifications for university admission. Motor delays were observed in 55% of the participants, speech development delays in 55%, and muscular hypotonia in 51%. Neurological symptoms were reported less frequently, including hearing impairment (13%), ataxia (9%), and visual impairment (7%). Notably, one female patient developed bilateral optic atrophy at the age of 9.5 years. By the age of 11 years, she was still able to follow movement with her right eye and perceive light and dark with both eyes (Grünert et al., 2013).

Studies have confirmed the presence of ocular disorders and accompanying visual functioning difficulties in patients with PA (Baumgartner et al., 2014; Forny et al., 2021; Williams et al., 2009). Optic neuropathy, as one of the more significant consequences, has been described in a small number of case studies. The pathophysiology of optic neuropathy in this context is not fully understood, though it is believed to result from the accumulation of toxic metabolites and oxidative stress (Arias et al., 2014). However, even in children without signs of optic neuropathy, ophthalmologic abnormalities such as nystagmus, ocular apraxia, esotropia, and similar conditions have been observed (Alvarez et al., 2015). Alvarez et al. (2015) emphasised the multisystemic consequences of PA, noting that visual status improved only modestly in some patients. Williams et al. (2009) and Arias et al. (2014), in their reports on individuals with Methylmalonic Acidemia and PA, refer to mitochondrial optic neuropathies, which are characterised by relatively symmetric visual acuity loss, early and marked colour perception loss, centrocecal visual field defects, and temporally pale optic discs. In some patients, altered visual evoked potentials (VEPs) have been noted, suggesting central afferent dysfunction and possible involvement of the optic pathways and/or central nervous system (CNS).

Visual impairment caused by CNS changes is referred to as cortical/cerebral visual impairment (CVI). CVI can result from prenatal and perinatal trauma such as asphyxia, respiratory distress syndrome, and various infections, and it is associated with conditions such as periventricular leukomalacia, intraventricular haemorrhage, seizures, genetic syndromes, metabolic disorders, and others (Boot et al., 2010; Pehere et al., 2018). As a result of neurodegenerative and metabolic conditions, CVI can occur in MELAS syndrome, ornithine transcarbamylase deficiency, Fabry disease, Leigh syndrome, and X-linked adrenoleukodystrophy, with changes in visual functioning becoming noticeable later in childhood (Ganesh & Rath, 2018). The manifestations of CVI vary, but certain behavioural characteristics are more common, depending on various contributing factors (Vučinić et al., 2025). Roman-Lantzy (2018) identified ten visual characteristics associated with cerebral visual impairment, distinct from oc-

ular causes of visual loss: light-gazing behaviours, lack of visual reflex responses, colour and familiar object preferences, reliance on a specific part of the visual field, better detection of moving versus static stimuli, difficulty recognising distant objects, delayed or slow visual responses, challenges perceiving complex visual scenes, and absence of reach guided by vision. Similar manifestations were reported in other studies (Altınbay & Taşkın, 2023; Lueck, 2010; Kran et al., 2024). Additional features include fluctuating visual performance, brief and unstable visual attention, navigating without visual reference (Fazzi et al., 2007), and simultanagnosia - difficulty perceiving more than one stimulus at a time and integrating visual information into a meaningful whole, resulting in an inability to perceive and comprehend environmental events (Bennett et al., 2020). Face or object recognition difficulties, visual field deficits, and oculomotor control problems impair visual scanning of spaces and materials (Chokron et al., 2021; Goodenough et al., 2021). Although ocular structures are often intact, CVI may be accompanied by reduced visual acuity and decreased contrast sensitivity (Vancleef et al., 2020). After detecting this type of visual impairment, it is essential to initiate vision (re)habilitation to support optimal visual development and functioning (Vučinić et al., 2019). Visual (re)habilitation for children with CVI relies on an individualised approach that includes structured activities and multisensory strategies to stimulate visual functions (Ganesh & Rath, 2018). Studies show that early vision (re)habilitation is particularly effective, providing favourable conditions for further development (Thapa, 2023; Sahli et al., 2021).

CASE REPORT

Initial contact with the family

The parents of Z. S., who was six years old at that time, sought professional support at the Counselling Centre of the Faculty of Special Education and Rehabilitation, University of Belgrade, aiming to receive consultation and conduct a functional vision assessment for their daughter. During the initial meeting with the parents, a comprehensive anamnesis was obtained, and an appointment for the functional vision assessment was scheduled in accordance with the child's and family's needs. Alongside verbally communicating the primary diagnosis - propionic acidaemia - the parents also provided additional medical documentation concerning her ophthalmological and neurological status.

VEP results indicated bilateral dysfunction of moderate severity. An ophthalmologic examination revealed sluggish pupillary reactions to light and pale-coloured optic nerve papillae. The girl had been prescribed corrective glasses with the following refractive values:

OD +2.50 / +1.00 ax90, **OS** +2.75 / +1.00 ax100.

She was undergoing anti-epileptic therapy. According to the parents, she had experienced multiple so-called "metabolic crises" that led to a deterioration in both motor and visual functioning, with episodes varying in intensity. The TEACH screening for CVI was administered (Screening list for children with a suspicion of Cerebral Visual Impairment Screening list 1; https://www.teachcvi.net/screening-tools), revealing the presence of 5 out of 6 indicators, which classifies the screening result as positive. The screening was conducted in collaboration with the parents following an extended period of visual behaviour observation. The primary challenge encountered during the assessment was evaluating the child's ability to recognise everyday objects, given the variability and fluctuation observed in visual functioning. In addition, the findings from the VEP, ophthalmological examination, and medical history further confirmed the presence of this type of visual impairment; however, an official diagnostic protocol for CVI had not yet been established in Serbia at that time. A few days after the initial visit, an increase in propionic acid levels was recorded in Z. S.'s blood, signifying another decline in metabolic status. The parents expressed concern regarding a deterioration in her vision, as the girl reported being able to see nothing but light. She was hospitalised, and several days after discharge, she regained the ability to recognise colours and some familiar household objects. Through ongoing observation, the parents noticed

that the time of day affected Z. S.'s functioning: more specifically, they observed that she used her vision more effectively in the afternoon than in the morning. A repeat ophthalmologic exam revealed no changes in visual status compared to previous findings, aside from slightly slower pupillary responses. In agreement with the parents, it was decided that the functional vision assessment would be conducted in the child's familiar environment at different times of the day, and all assessments were performed by the same professionals to ensure consistency.

First observation and assessment

The observation of visual behaviour was conducted during the morning hours. By observing spontaneous behaviour, it was noted that Z. S. relied heavily on auditory and tactile cues, even when navigating a well-known environment. Visual reflexes were present. Contrast sensitivity was assessed using the Hiding Heidi preferential looking test, and the girl responded only to the 100% contrast pattern. When presented with gold and red stimuli on a dark background, she was able to detect them at a distance of approximately 30 cm, reacting faster when the object was moving. Additional lighting of the object facilitated recognition and served as a motivating factor for exploring the environment. Visual tracking involved simultaneous movement of the head and eyes. Z. S. showed a preference for the colour yellow and used yellow elements that could move during play. The parents reported that she also preferred playing with yellow toys in her room. The visual field was assessed using the confrontation method. She exhibited delayed gaze orientation toward stimuli presented in the peripheral regions of the visual field. However, given the limited accuracy of this method and the inability to conduct more precise perimetric testing, potential visual field deficits could not be confirmed with certainty.

During the assessment, she asked questions and requested explanations about what was happening in her environment, clearly indicating her ability to perceive actions. When grasping objects, she demonstrated imprecision, often exploring the space around the object in order to determine its position more accurately and adjust her motor response accordingly.

Interventions:

Parents received advice on how to adapt the living and learning environment at home. It was suggested that they use dark coloured play and work surfaces, with the possibility to adjust the tilt of the desk. The rooms frequented by the girl were advised to be uncluttered, with toys neatly organised in boxes labelled with clear symbols or pictures so that Z. S. could easily identify where each item was located. Good lighting was recommended. Parents were advised to use slow-moving, high-contrast visual stimuli, as well as to allow sufficient response time in order to encourage gaze orientation and gradually improve visual responsiveness within the peripheral visual field.

For the purpose of providing additional lighting at the work desk, a lamp with a movable neck was recommended in order to allow light to be directed precisely to the desired observation target. For safe and motivated movement, it was proposed to mark thresholds with tape in a colour contrasting with the floor surface, as well as to mark the edges of items arranged in a row so that Z. S. could recognise them quickly while moving.

The parents implemented the above-mentioned instructions for the following two weeks. Several times daily, through every day and play activities, they encouraged the girl to visually notice adapted objects, reach for them, and recognise them. They encouraged her to move independently with verbal guidance and by pointing to the markings if Z. S. failed to notice them. When she could not locate an object during play, they used light guidance techniques to help her find it more easily.

Second observation

After two weeks, a new observation was conducted in the early evening. At first sight, Z. S. appeared significantly more cheerful and engaged in playing with items from a toy kitchen. The way she used objects during play indicated that her symbolic play skills were developed according to age-related expectations. She established and

maintained interaction by narrating events. She independently went to her room and located elements necessary to continue her play.

On a dark background, she was able to recognise colours, shapes, and some individual letters. On occasion, she needed to be within approximately 15-20 cm of an object in order to be able to identify it. When two objects were very similar, she had difficulty distinguishing them visually. Additional lighting continued to help her orient to and recognise targets. She had more difficulty noticing objects introduced into her visual field from the periphery and demonstrated a latency of up to 5 seconds in her response. Shifting her gaze from one object to another was challenging, though she successfully indicated the location of each object.

Repeated contrast sensitivity testing using the Hiding Heidi preferential test showed slight improvement: she could detect the 1.25% contrast pattern at a distance of 30-40 cm. She tracked a glittering object with head and eye movements, although circular tracking movements remained impaired. While grasping objects, she occasionally made errors requiring fine precision. During mobility, she had more difficulty noticing objects located in the lower visual field.

Interventions:

At this stage, in addition to previously provided advice for adapting materials and the environment, the parents were advised that they should involve the child in visual activities resembling vision training, adapted for CVI. The recommendations were tailored to the characteristics of CVI, including latency/delay in visual response, variable visual behaviour, difficulties detecting stimuli in visually complex environments, and preference for stimuli of certain colours.

Suggested activities included oculomotor exercises, figure-ground discrimination tasks, scanning exercises, detail extraction, and visual reasoning. Complementing the vision-specific activities, suggestions were also made for sensory integration exercises, particularly targeting proprioception and the vestibular system due to their importance for mobility and attention guidance. Additionally, tactile and auditory training elements were recommended to enrich visual information and facilitate spatial orientation and response speed.

It was agreed with the parents that they would implement these recommendations daily, while Z. S. would attend targeted sessions with a vision rehabilitation therapist three times a week.

Table 1. *Empirical measures across assessments*

Measure	First observation (morning)	Second observation (after 2 weeks, early evening)	After visual training (~ 1 month)
Contrast sensitivity (Hiding Heidi)	Detects only 100% contrast	Detects 1.25% contrast at 30-40 cm	Not reassessed
Response latency – peripheral visual field	Latency > 10 s	Latency up to 5 s	Latency up to ~ 3 s with adapted stimuli
Reaction to motion	Faster response to moving versus static targets	Maintained stronger response to motion	Not specifically quantified post-training
Gaze shifting (between two objects)	Not reliably assessed	Difficulties shifting gaze	More effective scanning strategies reported
Visual tracking (pursuit)	Head and eye combined; smooth pursuit limited	Partial improvement; circular tracking impaired	Not reported
Object recognition/ distance	Gold/red on dark background detected at ~ 30 cm	Recognition sometimes required approaching to within 15-20 cm; difficulty with highly similar objects	Able to handle new/partially adapted materials (reduced contrast)
Functional use of vision in activities	Relied on auditory/tactile cues; limited visual initiative	Independent exploration; locates items for play	Significant improvement in adapted spaces; outdoors/unfamiliar settings still require support

After one month of conducting the suggested activities in the adapted environment using appropriate materials, an evaluation was performed. Parental reports indicated significant improvement in Z. S.'s functioning within the adapted space, whereas she still sought support and guidance outdoors and in unfamiliar environments. Outside, she was able to locate familiar objects. Parents observed that visual functioning remained poorer in the morning hours, with improvement occurring later in the day when activities were most frequently conducted.

Regarding the visual training, faster responses were noted when adapted stimuli were presented (latency up to 3s), and Z. S. increasingly managed to cope with new and partially adapted materials (different background colour, reduced contrast). She independently used lighting when needed, and her improved and freer mobility in space enabled better exploration of objects around her. She employed more effective scanning strategies and performed equally well with both 2D and 3D materials. Despite the observed progress, visual fluctuation remained a central characteristic of her functioning, and variations in performance across different times of the day continued to persist.

In the following period, testing of fixation and scanning strategies using an eye gaze system was recommended to develop even more precise guidelines for functioning and learning.

CONCLUSION

Chronic and rare diseases can profoundly affect family life and cause psychosocial stress among parents and other family members. Daily pressures, uncertainty, financial burden, and constant concern for the child's future can add further strain on parents and influence the emotional well-being and status of siblings. From the parents' perspective, difficulties in the child's interaction with peers represents another common challenge (Grünert et al., 2013).

As soon as PA is suspected, the patient should be referred to a specialised institution in order to determine the necessary interventions and specific therapies required. Caring for a child with a chronic, potentially life-threatening condition necessitates a significant role of parents throughout the entire treatment and (re)habilitation process. Early diagnosis of PA and timely treatment are essential and can provide significant long-term benefits. In addition to medical care, the child requires ongoing monitoring by vision rehabilitation specialists. There is a lack of large-scale studies investigating the presence of CVI in patients with PA; therefore, evidence from case series or smaller studies must be carefully examined and made available to the scientific community and families. Case studies involving CVI treatment in children with PA are particularly rare. In our case study of Z. S., visual functioning difficulties were identified through observation of her visual behaviour, parental anamnesis, and screening for CVI, confirming the presence of CVI characteristics. Based on the information collected, a proposal was developed for necessary adaptations and a visual training programme. Positive effects were observed within the first month of implementation. Although PA is a condition with an unpredictable outcome, it is essential to conduct continuous evaluations and monitor visual development and functioning in affected children to improve their quality of life and ensure adequate education and daily functioning.

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