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Sección
Artículos de Revisión

Interdisciplinary Rehabilitation in a Patient with an ACY1 Mutation and Post-Anoxic Encephalopathy Secondary to Epiglottitis: Case Report

Rehabilitación interdisciplinaria en paciente con mutación ACY1 y encefalopatía postanóxica secundaria a epiglotitis. Reporte de caso

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Abstract

Introduction: Aminoacylase 1 (ACY1) deficiency is a rare inborn metabolic disorder associated with variable neurological impairment. Acute epiglottitis may trigger severe hypoxia and post-anoxic encephalopathy. This case describes the functional impact of an interdisciplinary neurorehabilitation program incorporating visual therapy to enhance sensorimotor reorganization. **Case Presentation**: A 22-year-old male with homozygous ACY1 mutation, psychomotor delay, hypotonia, sensorineural hearing loss, and multiple comorbidities developed airway obstruction secondary to epiglottitis, resulting in anoxic injury and coma. He subsequently exhibited dysphagia, spasticity, dystonia, ataxia, disorganized body schema, and visuospatial and oculomotor deficits (exotropia, convergence insufficiency, monocular suppression). He was fully dependent for basic activities and showed severely reduced FIM, PASS, and FAC scores. **Discussion**: An intensive program combining physiotherapy, occupational therapy, speech therapy, and specialized visual therapy was delivered. A Fresnel prism and structured oculomotor, binocular integration, and spatial-perception exercises facilitated improvements in postural control, motor precision, and visual stability. Dystonia decreased substantially, and the patient progressed toward assisted gait. Follow-up assessments demonstrated clinically meaningful gains in FIM, PASS, and FAC scores. **Conclusions**: Integrating visual therapy into interdisciplinary neurorehabilitation may enhance visuospatial integration and motor modulation in complex post-anoxic encephalopathies linked to rare metabolic disorders. This case supports its role as a valuable adjunctive intervention.

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Keywords: neurological rehabilitation; brain diseases; physical therapy; optometry; vision disorders.

Resumen

Introducción: La deficiencia de aminoacilasa 1 (ACY1) es un trastorno metabólico congénito poco frecuente asociado a una afectación neurológica variable. La epiglotitis aguda puede desencadenar hipoxia grave y encefalopatía postanóxica. Este caso describe el impacto funcional de un programa interdisciplinar de neurorrehabilitación que incorpora terapia visual para mejorar la reorganización sensoriomotora. **Presentación del caso**: Varón de 22 años con mutación homocigota en ACY1, retraso psicomotor, hipotonía, hipoacusia neurosensorial y múltiples comorbilidades, que desarrolló obstrucción de la vía aérea secundaria a epiglotitis, lo que derivó en lesión anóxica y coma. Posteriormente presentó disfagia, espasticidad, distonía, ataxia, esquema corporal desorganizado y déficits visuoespaciales y oculomotores (exotropía, insuficiencia de convergencia, supresión monocular). Era completamente dependiente para las actividades básicas y presentaba puntuaciones gravemente reducidas en las escalas FIM, PASS y FAC. **Discusión**: Se aplicó un programa intensivo que combinó fisioterapia, terapia ocupacional, logopedia y terapia visual especializada. La utilización de un prisma de Fresnel y de ejercicios estructurados de oculomotricidad, integración binocular y percepción espacial facilitó mejoras en el control postural, la precisión motora y la estabilidad visual. La distonía disminuyó de forma significativa y el paciente progresó hacia la marcha asistida. Las evaluaciones de seguimiento mostraron mejoras clínicamente relevantes en las puntuaciones FIM, PASS y FAC. **Conclusiones**: La integración de la terapia visual en la neurorrehabilitación interdisciplinar puede mejorar la integración visuoespacial y la modulación motora en encefalopatías postanóxicas complejas asociadas a trastornos metabólicos raros. Este caso respalda su papel como una intervención complementaria valiosa.

Palabras clave: rehabilitación neurológica; enfermedades cerebrales; fisioterapia; optometría; trastornos de la visiónsináptica; dieta ultraporcesada.

1. INTRODUCTION

Aminoacylase 1 (ACY1) deficiency is a rare congenital metabolic disorder caused by homozygous mutations in the ACY1 gene, located on chromosome 3p21.2. It follows an autosomal recessive inheritance pattern and is classified as an organic aciduria, characterized by the accumulation of N-acetylated amino acids in the urine [1, 2]. Clinical presentation is highly variable, most commonly including encephalopathy, generalized muscle weakness, and hypotonia. Epiglottitis, a supraglottic inflammation most frequently caused by Haemophilus influenzae type b, may progress to severe airway obstruction and, in critical cases, result in anoxic encephalopathy [3, 4].

Scientific evidence regarding motor and functional recovery after post-anoxic encephalopathy remains limited, particularly in patients with rare metabolic disorders and when complementary approaches such as visual therapy are incorporated into neurorehabilitation programs. There is a lack of documented cases describing interdisciplinary neurorehabilitation strategies in patients with ACY1 deficiency who develop post-anoxic encephalopathy secondary to epiglottitis.

ACY1 deficiency has been associated with heterogeneous neurological features, including psychomotor delay, seizures, hypotonia, cerebellar or corpus callosum abnormalities, and, in less frequent cases, craniofacial and sensory alterations [1, 2]. Recent research suggests that ACY1 may also play a role in inflammatory regulation, although this relationship has not yet been fully established [5].

Epiglottitis remains a potentially life-threatening condition due to the risk of acute airway obstruction, which may lead to hypoxic-ischemic brain injury and diffuse encephalopathy [3, 4]. Post-anoxic encephalopathy often results in significant cognitive, motor, and sensory impairments that compromise autonomy and functional independence.

Neurorehabilitation aims to restore or compensate for neurological deficits following central nervous system injury. However, evidence regarding the effectiveness of complementary therapies such as visual therapy in post-anoxic brain injury is still limited. Visual therapy is a neurophysiological stimulation approach designed to improve visual acuity, ocular motility, binocular vision, spatial orientation, and sensory integration. In patients with acquired brain injury, visual-perceptual deficits may significantly affect postural control, coordination, and motor planning, thereby limiting functional recovery [6, 7].

The purpose of this case report is to describe the interdisciplinary neurorehabilitation process of a young adult patient with ACY1 deficiency who developed post-anoxic encephalopathy secondary to epiglottitis. Specifically, this report aims to analyze the contribution of visual therapy within a comprehensive rehabilitation program and its potential impact on postural control, coordination, and functional recovery. It is hypothesized that the integration of visual therapy may positively influence motor performance and overall functional outcomes in this clinical context.

2. OBJECTIVE

The objective of this case report is to describe the interdisciplinary neurorehabilitation process of a patient with brain injury secondary to post-anoxic encephalopathy, with particular emphasis on the integration of visual therapy within a physiotherapy-based motor rehabilitation program.

The justification for reporting this case lies in the limited evidence available regarding the role of visual therapy as an adjunct intervention in motor neurorehabilitation, especially in patients with complex neurological conditions and rare metabolic disorders. Although visual-perceptual deficits are common following acquired brain injury and may significantly interfere with postural control, coordination, and functional performance, their systematic integration into rehabilitation protocols remains insufficiently documented. By detailing the clinical progression and functional outcomes observed in this patient, this report aims to contribute clinically relevant evidence to the existing literature and to support further investigation into interdisciplinary approaches targeting sensorimotor reorganization and functional recovery.

3. METHOD

3.1 Description of the Context and the Case

This study follows a longitudinal pre–post interventional case report design. Patient A is a 22-year-old individual diagnosed with organic aciduria secondary to aminoacylase 1 deficiency caused by a homozygous mutation in the ACY1 gene. During early development, Patient A presented psychomotor delay and generalized hypotonia. In adolescence, bilateral sensorineural hearing loss was diagnosed, and a cochlear implant was placed. Additional comorbidities documented over time included arterial hypertension, intestinal obstruction requiring multiple surgical interventions, concentric hypertrophic cardiomyopathy of the left ventricle, and right-sided pneumonia secondary to SARS-CoV-2 infection.

Patient A was admitted to the emergency department in a comatose state (Glasgow Coma Scale score: 6), requiring orotracheal intubation. Following recovery from coma, neurological sequelae were observed, including dysphagia requiring gastrostomy tube placement, predominantly upper-limb spasticity, and myoclonus treated with anticonvulsant medication (levetiracetam, oral administration, dosage adjusted according to body weight and neurological response).

From hospital admission to discharge, one year and four months elapsed. Four months after discharge, Patient A was admitted to our neurorehabilitation center. Therefore, the initial evaluation was conducted one year and eight months after the brain injury.

Written informed consent for publication was obtained from the patient's legal guardians. All procedures were conducted in accordance with ethical standards for clinical

case reporting and in compliance with institutional guidelines. No personally identifiable information is disclosed.

3.2 Materials

The materials used during assessment and intervention included:

- Standardized functional assessment scales (FIM, PASS, FAC).
- Optometric assessment tools, including visual acuity charts and the Worth four-dot test.
- Fresnel prism prescribed for the left eye.
- Neurorehabilitation equipment for physiotherapy, including balance training devices and postural control materials.
- Visual therapy materials designed to stimulate ocular motility, convergence, fixation, and spatial perception.

All materials were selected according to clinical indication and patient-specific needs.

3.3 Instruments

The following standardized assessment instruments were administered pre- and post-intervention:

- Functional Independence Measure (FIM) [8], used to assess functional independence in activities of daily living.
- Postural Assessment Scale for Stroke (PASS) [9], used to evaluate postural control in lying, sitting, and standing positions.
- Functional Ambulation Categories (FAC) [10], used to classify gait ability.
- Worth four-dot test, used to assess binocular vision and sensory fusion.

3.4 Procedures

3.4.1 Assessment

At baseline, a comprehensive physiotherapeutic evaluation was conducted. Initial scores were:

- FIM: 32
- PASS: Mobility 10; Balance 2
- FAC: 0

Clinical observation revealed significant proprioceptive alteration, disorganized body schema, ataxic movement patterns, dystonia, and complete dependence for transfers and activities of daily living. Communication was limited to yes/no head movements.

Due to observed impairments in ocular tracking, distance estimation, and movement precision, Patient A was referred for optometric evaluation. The assessment revealed reduced distance visual acuity (more pronounced in the left eye), deficient ocular tracking precision, exotropia at near and far distances, convergence disorder, and suppression of the left eye on the Worth test.

3.4.2 Intervention

An intensive multidisciplinary neurorehabilitation program was implemented, consisting of:

- Physiotherapy: 4 hours/week
- Occupational therapy: 2 hours/week
- Speech therapy: 2 hours/week
- Visual therapy: 1 hour/week

Physiotherapy initially focused on body awareness, spatial orientation, postural control, and movement organization. However, excessive involuntary movements limited motor control and sensory integration.

Following interdisciplinary discussion, a Fresnel prism was prescribed for the left eye, and a structured visual therapy program was initiated with weekly sessions targeting ocular motility, convergence, fixation stability, and visuospatial integration.

The total intervention period lasted 12 months. All sessions were individualized and progressively adapted according to Patient A's tolerance, motor control, and learning rate.

3.4.3 Post-intervention Assessment

After 12 months of intervention, the same standardized scales were re-administered:

- FIM: 67
- PASS: Mobility 17; Balance 8
- FAC: 2

Clinical improvements included reduction of dystonia, enhanced postural control, improved coordination, initiation of assisted gait, and increased independence in activities of daily living.

4. RESULTS

4.1 Quantitative Outcomes

After 12 months of multidisciplinary intervention, improvements were observed in all standardized functional assessment scales.

The Functional Independence Measure (FIM) score increased from 32 at baseline to 67 at post-intervention (Δ +35 points).

On the Postural Assessment Scale for Stroke (PASS), mobility improved from 10 to 17 (Δ +7), and balance improved from 2 to 8 (Δ +6).

The Functional Ambulation Categories (FAC) score increased from 0 to 2, indicating progression from non-functional ambulation to assisted ambulation. Detailed results are presented in Table 1.

Table 1. Functional assessment results at baseline and post-intervention

Scale	Baseline	Post-intervention	Change
FIM	32	67	+35
PASS mobility	10	17	+7
PASS Balance	2	8	+6
FAC	0	2	+2

Source: Own elaboration

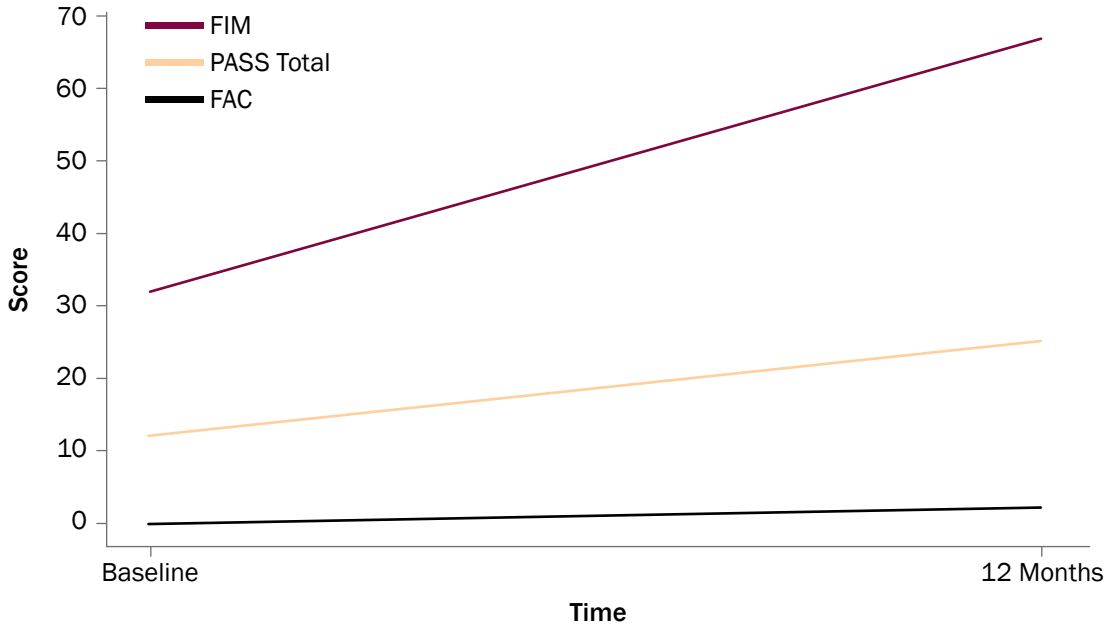
4.2 Clinical Motor Outcomes

From a clinical perspective, a marked reduction in dystonic movements was observed. At baseline, dystonia was present during most voluntary motor tasks, whereas post-intervention it appeared only occasionally during activities requiring high motor precision or increased muscular effort.

Improvements were observed in static balance (sitting and standing), dynamic balance, body coordination, and motor organization in space. Patient A demonstrated increased ability to execute movements in response to verbal commands with improved precision and reduced motor disorganization (see Figure 1).

Assisted gait was initiated during the intervention period. Additionally, partial gains in independence were observed in basic activities of daily living, particularly in transfers and postural maintenance.

Figure 1. Temporal evolution of functional outcomes



Source: Own elaboration

4.3 Visual Outcomes

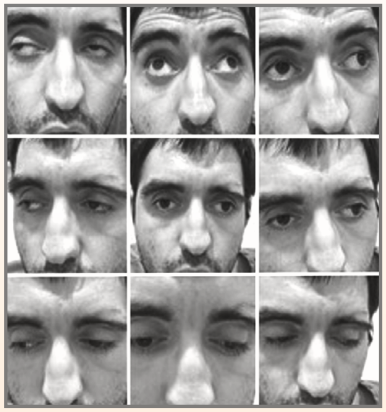
Optometric follow-up assessment demonstrated improved ocular mobility and control across the nine gaze positions. Convergence capacity showed functional improvement, and suppression of the left eye was reduced under therapeutic conditions.

A comparative visual record of ocular motility before and after intervention is presented in Figure 2.

Figure 2. Evolution of ocular mobility

Pre intervention

Post intervention



5. DISCUSSION

This case illustrates the complexity of neurorehabilitation in a patient with brain injury secondary to cerebral hypoxia, particularly in the context of a rare metabolic disorder such as aminoacylase 1 deficiency. Although the literature on this condition is limited, the neurological sequelae observed in Patient A are consistent with those described in hypoxic-ischemic brain injury.

The quantitative improvements documented in the Results section, including increases in FIM, PASS, and FAC scores, indicate relevant functional gains in independence, postural control, and assisted ambulation. These findings are aligned with the initial hypothesis that integrating visual therapy into a motor neurorehabilitation program may contribute to sensorimotor reorganization and functional recovery.

From a physiotherapeutic perspective, environmental perception and body awareness are essential components of motor planning and execution. The addition of visual therapy allowed specific visuospatial and oculomotor impairments, such as altered spatial perception, exotropia, and convergence deficits, to be addressed within the rehabilitation process. Prism adaptation has been described in neurological contexts as a strategy capable of modifying spatial perception and facilitating cortical reorganization after brain injury. Visual therapy has also been recognized as a complementary approach to improve functional and motor performance in patients with sensory or visuospatial integration deficits [11].

In the present case, a noticeable change in motor organization was observed after prism prescription, accompanied by a reduction in dystonic and ataxic movements during functional tasks. While a causal relationship cannot be established in a single-case design, the temporal association between visual intervention and motor improvement suggests a possible interaction between visual input and motor control mechanisms.

Through interdisciplinary collaboration, previously undetected functional visual deficits were identified, which may have influenced the patient's interaction with the environment and motor responses. Progressive improvements in standardized functional scales (FIM, FAC, PASS) support the relevance of a coordinated intervention strategy in this clinical context.

However, the evidence supporting the role of visual therapy within motor neurorehabilitation remains limited and heterogeneous [6, 12, 13]. This report describes a single patient who underwent simultaneous multidisciplinary treatment, making it impossible to isolate the specific contribution of visual therapy. Additionally, spontaneous neurological recovery over time cannot be ruled out. Therefore, these findings should be interpreted with caution.

Further studies including larger samples and longer follow-up periods are necessary to better clarify the potential contribution of visual therapy within neurorehabilitation programs and to deepen understanding of how visual function may influence motor recovery and patient autonomy [6, 12, 13].

6. CONCLUSION

This case illustrates the complexity of neurorehabilitation in post-anoxic encephalopathy associated with aminoacylase 1 deficiency and highlights the potential relevance of an interdisciplinary approach. Functional improvements in postural control, coordination, and independence were observed following combined physiotherapy and optometric intervention. The integration of visual therapy enabled the identification and management of visuospatial and oculomotor deficits that may influence motor performance. Although causality cannot be established in a single-case design, these findings support the value of comprehensive assessment and interdisciplinary collaboration in neurological rehabilitation. Further research is warranted to better clarify the contribution of visual therapy to motor and functional recovery.

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