VISUAL AND NEUROMOTOR REHABILITATION OUTCOMES IN A CHILD WITH A STXBP1 AUTOSOMAL DOMINANT DEVELOPMENTAL AND EPILEPTIC ENCEPHALOPATHY, TYPE 4 WITH AXIAL HYPOTONIA, GLOBAL DEVELOPMENTAL DELAY, AND ADHD: A CASE STUDY

Rabindra Kumar Pandey M.Optom, FOVDRA-USA, FAAO, MCOptom-UK,(PhD),Priya Pandey M.Optom, FCSO-USA, MCOptomUK,(PhD) Debesh Mitra, B.Optom, PGDOVS

Contact US: Caringvisiontherapy@gmail.com

Introduction

Developmental and Epileptic Encephalopathy-4 (DEE-4) is a severe neurological disorder of infancy classified under early infantile epileptic encephalopathies. It is most commonly associated with pathogenic variants in the STXBP1 gene, which plays a critical role in synaptic neurotransmission. The condition typically presents with early-onset tonic seizures that are progressive and often refractory to treatment, contributing to significant encephalopathic impairment.

Children with DEE-4 exhibit profound psychomotor developmental impairment. Developmental delay is global, affecting motor, cognitive, and speech domains. Major motor features include spastic quadriplegia, decreased muscle tone involving all four limbs, difficulty maintaining posture and balance, and abnormal posture with persistence of primitive reflexes. Overall motor control and coordination are significantly compromised.

Visual efficiency is markedly reduced due to the combined effects of poor visual acuity, impaired visual processing, reduced attention span, motor and postural instability, and cognitive impairment. These deficits collectively result in difficulty with reading readiness, poor mobility vision, reduced learning through visual input, and increased dependence on alternative sensory modalities such as touch and sound for environmental interaction.

General Clinical Findings

The patient presents with a complex neurodevelopmental disorder characterized by multisystem involvement. Musculoskeletal findings included abnormal calf and thumb morphology, Achilles tendon contracture, genu recurvatum, foot dorsiflexor weakness, and generalized (predominantly proximal) muscle weakness with axial and global hypotonia, intermittent stiffness, and spasticity.

There was global developmental delay with significant motor delay, including delayed standing and ambulation. Gait was broad-based and ataxic with imbalance and postural instability, leading to functional limitations and easy fatigability.

Neurological examination revealed apraxia, dysarthria, tremor, myoclonus, involuntary movements, and persistence of primitive reflexes. Behavioral assessment showed delayed social milestones, reduced eye contact, hyperactivity, and marked speech delay.

Neuroimaging demonstrated cerebral and periventricular white matter abnormalities, ventricular dilatation, cerebellar atrophy, and a thin corpus callosum. EEG showed multifocal epileptiform discharges, and clinically the patient exhibited multiple seizure types with associated sleep disturbance.

Overall, the findings are consistent with a severe neurodevelopmental condition with structural brain abnormalities, multifocal epilepsy, motor dysfunction, and global developmental impairment.

Case Summary & Methodology

Patient Demographics & Background

Parameter	Details
Age at Presentation	6 years 4 months
Gender	Male
Diagnosis	STXBP1-related autosomal dominant DEE (Type-4)
Associated Conditions	Axial hypotonia, ADHD
Primary Concerns	Poor fixation, difficulty tracking, reduced eye contact

Birth & Medical History

Category	Findings
Birth History	Full-term, Caesarean delivery
NICU Admission	None
Neonatal Jaundice	Absent
Prenatal/Postnatal Complications	None reported
Ocular History	No prior surgery; no previous refractive correction

Current Medications

Medication	Indication
Oxerol	Seizure control
Lacosamide	Seizure control
Amantrel 10 mg	Tremor management

Clinical Examination

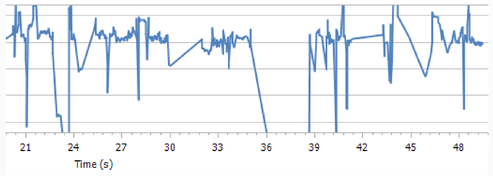
A. Visual Efficiency

Parameter	Pre VT	Post VT
Fixation	Poor, unstable	
Saccades & Pursuits	Poor; excessive head/body movement	
Visual Acuity RE	6/18	6/6
Visual Acuity LE	6/12	6/6
Objective Refraction (RE)	+1.25 / −2.25 × 10	
Objective Refraction (LE)	+1.00 / −1.00 × 180	
Near Point of Convergence	>25cm	10cm
Stereoacuity	<800 seconds of arc	40 seconds of arc

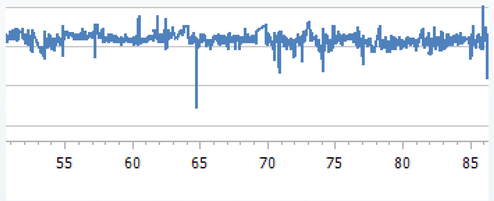
B. Primitive Reflex Assessment

Reflex	Pre VT Grade	Post VT Grade
Palmar Reflex	2	0
Babinski Reflex	4	0
Rooting Reflex	4	0
Supine Moro Reflex	3	0
Tonic Labyrinthine Reflex (TLR)	4	2
Asymmetrical Tonic Neck Reflex (ATNR)	3	2

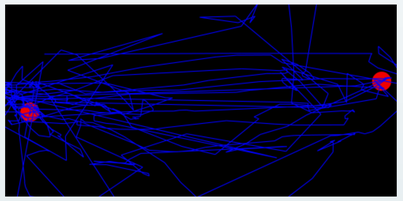
Methodology



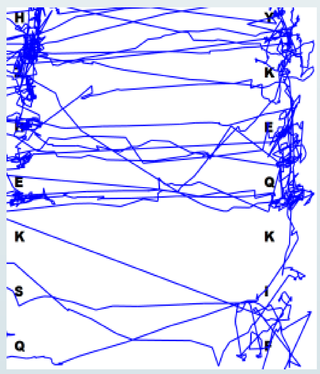
Pre-VT horizontal disparity



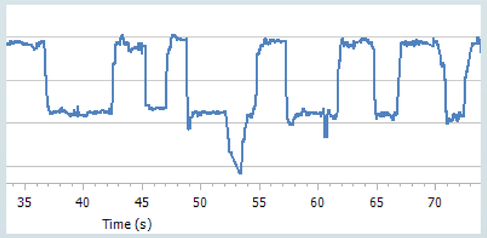
Post-VT horizontal disparity



Pre-VT saccades



Post-VT letter saccades



Post-VT letter saccades eye jump

Note- Pre-VT letter saccades were not recordable

Results

STXBP1-related developmental and epileptic encephalopathy is characterized by severe neurodevelopmental impairment, often accompanied by visual efficiency deficits, delayed motor integration, and attentional challenges. Visual dysfunction in such children is frequently underrecognized, despite its critical role in cognitive development, motor planning, and social interaction.

This case highlights the interrelationship between visual efficiency, primitive reflex integration, and neurodevelopmental maturity. The presence of multiple retained primitive reflexes likely contributed to impaired ocular motor control, poor fixation stability, and compromised binocular function. A structured, phase-wise vision therapy program combined with reflex integration therapy resulted in significant improvements across visual acuity, binocular vision, stereoacuity, convergence ability, and visual perceptual processing.

The progressive integration of primitive reflexes paralleled improvements in visual motor control and perceptual skills, suggesting a neuroplastic response to targeted multisensory intervention. Importantly, the child demonstrated measurable functional gains despite the underlying genetic encephalopathy, emphasizing the potential role of vision therapy as a supportive rehabilitative modality in complex neurodevelopmental disorders.

Conclusion

This case demonstrates that individualized vision therapy combined with reflex integration therapy can lead to meaningful improvements in visual function and perceptual skills in a child with STXBP1-related developmental and epileptic encephalopathy. Early identification of visual efficiency deficits and retained primitive reflexes, followed by a structured, multidisciplinary therapeutic approach, may enhance functional visual outcomes even in genetically mediated neurodevelopmental conditions. Vision rehabilitation should therefore be considered an integral component of holistic neurodevelopmental management in children with developmental epileptic encephalopathies.

Reference