COEXISTING WEST SYNDROME, EPILEPTIFORM ENCEPHALOPATHY, OPEN LIP SCHIZOENCEPHALY AND SUSPECTED MITOCHONDRIAL DISEASE IN AN INFANT: A CASE REPORT

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ABSTRACT

Global developmental delay in infancy can arise from a wide range of neurological and structural brain disorders. Early recognition is important because multiple conditions may coexist and influence both clinical presentation and long-term outcome. We report the case of an infant who presented with developmental delay, poor attainment of age-appropriate milestones and recurrent epileptic spasms. Clinical evaluation showed features suggestive of epileptic encephalopathy and further assessment supported a diagnosis of West syndrome. Neuroimaging revealed open-lip schizencephaly, indicating a structural abnormality of brain development. Additional clinical and laboratory findings were suggestive of underlying mitochondrial disease. The coexistence of epileptiform encephalopathy, West syndrome, open-lip schizencephaly and suspected mitochondrial dysfunction created a complex neurological picture with significant developmental impairment. This case highlights the importance of careful neurological examination, neuroimaging, and metabolic evaluation in infants presenting with developmental delay and early-onset seizures. Early diagnosis helps guide management, supports counselling of caregivers and may improve long-term developmental care planning.

KEYWORDS

Global developmental delay, epileptiform encephalopathy, West syndrome, mitochondrial disease, open-lip schizencephaly, VEEG, neuroimaging.

INTRODCUTION

Global developmental delay refers to significant delay in achieving developmental milestones in two or more domains, including motor, language, cognitive, social, and adaptive functioning. It is a common neurological concern in infancy and early childhood and may result from genetic, metabolic, structural, or acquired causes. Careful evaluation is important because early developmental delay can be the first sign of an underlying disorder affecting brain development[1].

Epileptiform encephalopathy is a group of severe neurological conditions in which repeated epileptic activity contributes to developmental slowing or loss of previously acquired skills. Among these disorders, West syndrome usually presents during infancy with epileptic spasms, developmental regression or delay, and characteristic electroencephalographic abnormalities. Early recognition is important because ongoing seizures may further affect neurodevelopmental outcome[2].

Open-lip schizencephaly is a rare congenital malformation of cortical development characterised by a cleft extending from the surface of the brain to the ventricular system. The clinical presentation can vary widely and may include developmental delay, motor impairment, and seizures[3].

Mitochondrial disease represents a group of disorders caused by impaired cellular energy production. In infants and children, these conditions may involve the central nervous system and present with developmental delay, hypotonia, seizures, and progressive neurological dysfunction[4].

The causes of global developmental delay with associated West syndrome, Epileptic encephalopathy, Open-lip schizencephaly, and suspected Mitochondrial disease are often varied and may involve more than one underlying factor. Important causes include abnormalities in brain development during pregnancy, genetic or chromosomal changes, metabolic disorders, mitochondrial dysfunction, infections affecting the fetus, reduced oxygen supply around birth, intracranial bleeding, and other forms of early brain injury[5].

Risk factors may include maternal infections during pregnancy, exposure to harmful substances, poor antenatal health, prematurity, birth complications, family history of neurological or genetic disorders, and structural abnormalities of the developing brain. These factors can interfere with normal brain maturation and may lead to developmental delay, seizures, and other neurological problems in infancy[6].

Infants with global developmental delay associated with West syndrome, Epileptic encephalopathy, Open-lip schizencephaly, and suspected Mitochondrial disease may show delayed achievement of developmental milestones such as poor head control, delayed sitting, reduced social interaction, and limited speech or vocalisation[7].

Common symptoms include recurrent seizures or infantile spasms, episodes of sudden body stiffening or jerking, poor feeding, irritability, abnormal eye movements, reduced alertness, and loss of previously acquired skills. Some infants may also have low muscle tone or muscle stiffness, weakness, poor coordination, and difficulty with movement. Depending on the extent of brain involvement, symptoms can vary in severity and may affect growth, behaviour, and overall neurological development[8].

Global developmental delay associated with West syndrome, Epileptic encephalopathy, Open-lip schizencephaly, and suspected Mitochondrial disease can lead to several long-term complications. Persistent seizures may interfere with brain development and can worsen cognitive, motor, and language abilities. Affected children may have learning difficulties, behavioural problems, movement disorders, muscle weakness or stiffness, feeding difficulties, poor growth, and problems with balance and coordination[9].

Some may develop recurrent hospital admissions because of uncontrolled seizures, infections, or metabolic stress. In more severe cases, there may be progressive neurological impairment, loss of previously acquired skills, and lasting limitations in daily functioning and quality of life[10].

The diagnosis of global developmental delay associated with West syndrome, Epileptic encephalopathy, Open-lip schizencephaly, and suspected Mitochondrial disease is based on detailed clinical assessment, developmental history, and neurological examination. Doctors assess delayed developmental milestones, seizure pattern, muscle

tone and any loss of previously acquired skills. Electroencephalography (EEG) helps identify abnormal electrical brain activity and can support the diagnosis of West syndrome and epileptic encephalopathy[11].

Brain imaging, especially magnetic resonance imaging (MRI), is important to detect structural abnormalities such as open-lip schizencephaly. Metabolic investigations, blood tests, lactate levels, genetic testing, and other supportive laboratory studies may also be done when mitochondrial disease is suspected. Combining clinical findings with neurophysiological, imaging, and metabolic evaluation helps establish the diagnosis and guide management[12].

Management of global developmental delay associated with West syndrome, Epileptic encephalopathy, Open-lip schizencephaly, and suspected Mitochondrial disease usually requires a multidisciplinary approach. Treatment mainly focuses on controlling seizures, supporting development, and managing associated neurological problems. Anti-seizure medicines may be used to reduce infantile spasms and recurrent seizures. Developmental support through physiotherapy, occupational therapy, speech therapy, and early intervention programmes can help improve motor, communication, and daily functioning skills. Nutritional support and regular monitoring of growth, feeding, and overall health are also important[13].

When mitochondrial disease is suspected, metabolic evaluation, supportive care, and avoidance of factors that may worsen metabolic stress are considered. Regular follow-up with pediatricians, neurologists and rehabilitation specialists helps to monitor progress and adjust treatment according to the child's needs[14].

The coexistence of global developmental delay, epileptiform encephalopathy, west syndrome, open-lip schizencephaly and suspected mitochondrial disease is uncommon and creates a complex diagnostic and management challenge. This case highlights the importance of detailed clinical assessment, neuroimaging, and metabolic evaluation in infants presenting with developmental delay and early-onset seizures[15].

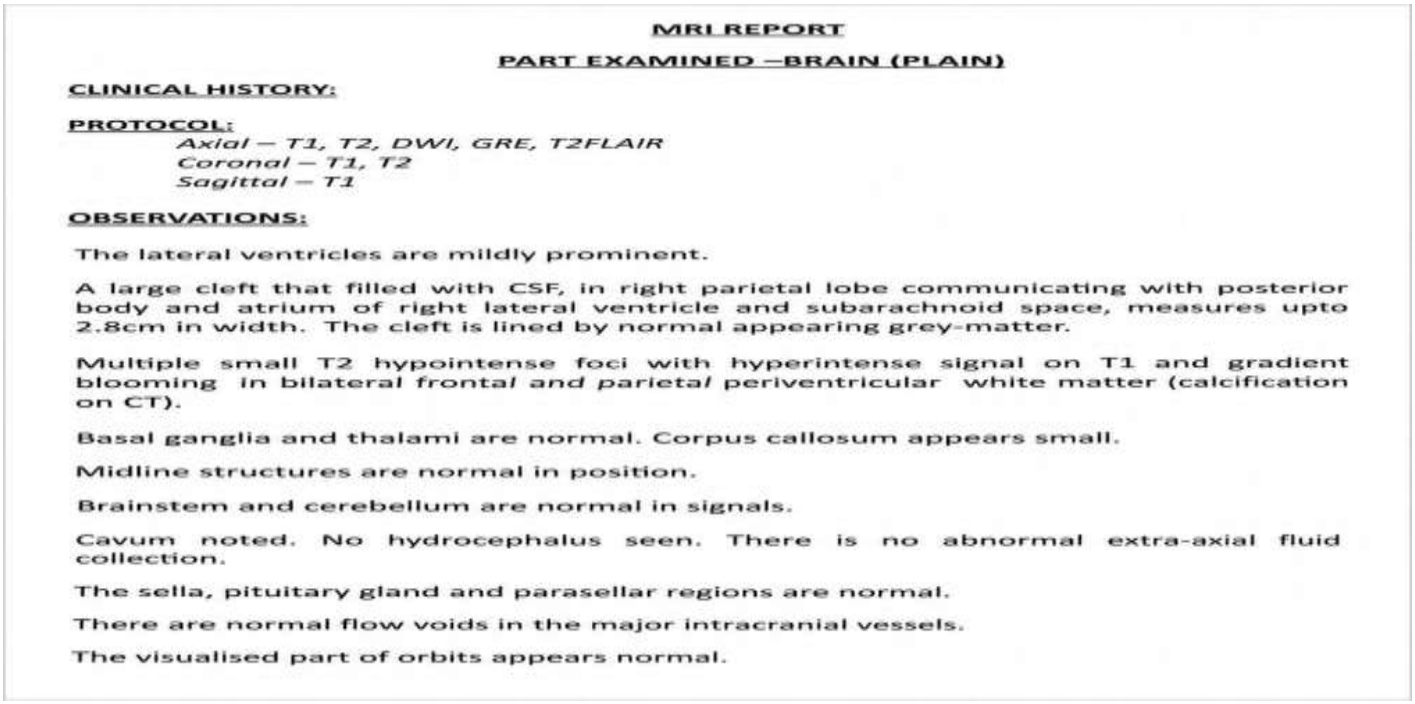
CASE PRESENTATION

A 1 year old male child was presented with multiple episodes of vomiting since 3 days, history of fever and fall since 3 days.He has a past medical history of hypothyroidism, at the 8th month of age he was detected with mitochondrial dias, west syndrome, global developmental delay and epileptiform ecephalopathy. He had an seizure episode at two and a half months of age.He has a past medication history of SYP. SODIUM VALPROATE (200mg/5ml) P/O BD, SYP.CLOBAZAM 2.5mg P/O HS, TAB.PHENOBARBITONE 30mg ½ HS and TAB.LEVOTHYROXINE SODIUM 25 mcg P/O OD.The laboratory investigation report showed elevated parameters are PCV, WBC count, Lymphocytes, CRP and declined parameters were polymorphs.

Figure 1:Affected infant

MRI brain showed open lip schizencephaly in the right parietal region, multiple small nodular calcifications in bilateral, frontal and parietal region and also showed a possibility of intra TORCH infection. The MRI scan revealed open-lip schizencephaly in the right parietal region, characterized by a cerebrospinal fluid-filled cleft extending from the cortical surface to the lateral ventricle, suggesting a congenital neuronal migration disorder affecting the right parietal lobe. Additionally, multiple small nodular calcifications were noted in the bilateral frontal and parietal periventricular regions, which are suggestive of a previous intrauterine infectious or inflammatory insult. These imaging findings raised the possibility of intrauterine TORCH infection, particularly congenital cytomegalovirus infection, which is commonly associated with periventricular calcifications and structural brain abnormalities. The combination of these abnormalities may contribute to severe neurological manifestations including developmental delay, epileptic spasms, and epileptic encephalopathy.

CT Brain showed CSF filled cleft extending from the cortical surface to the lateral ventricles lined by grey matter, right sided open cleft lip schizencephaly and bilateral periventricular coarse calcifications secondary to congenital cytomegalovirus infection.



MRI REPORT

PART EXAMINED –BRAIN (PLAIN)

CLINICAL HISTORY:

PROTOCOL:

Axial – T1, T2, DWI, GRE, T2FLAIR
Coronal – T1, T2
Sagittal – T1

OBSERVATIONS:

The lateral ventricles are mildly prominent.

A large cleft that filled with CSF, in right parietal lobe communicating with posterior body and atrium of right lateral ventricle and subarachnoid space, measures upto 2.8cm in width. The cleft is lined by normal appearing grey-matter.

Multiple small T2 hypointense foci with hyperintense signal on T1 and gradient blooming in bilateral frontal and parietal periventricular white matter (calcification on CT).

Basal ganglia and thalami are normal. Corpus callosum appears small.

Midline structures are normal in position.

Brainstem and cerebellum are normal in signals.

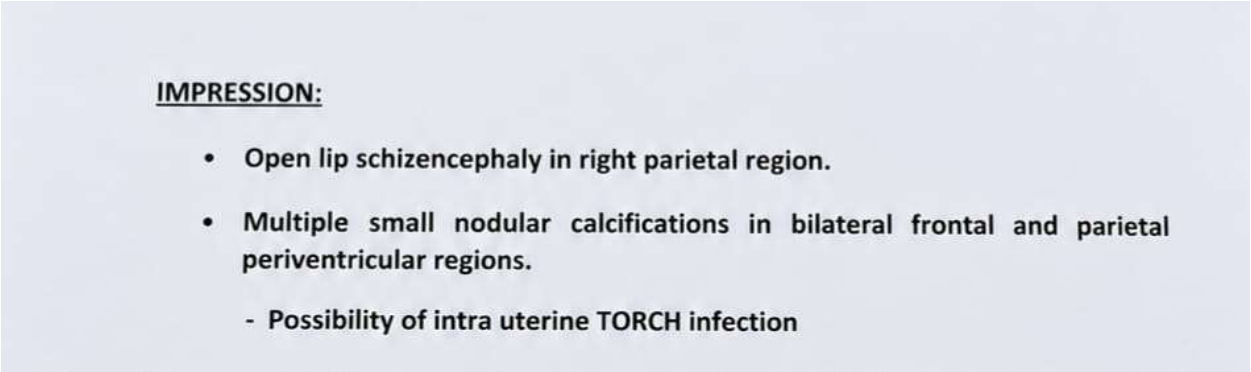
Cavum noted. No hydrocephalus seen. There is no abnormal extra-axial fluid collection.

The sella, pituitary gland and parasellar regions are normal.

There are normal flow voids in the major intracranial vessels.

The visualised part of orbits appears normal.

Figure 2: MRI Brain Report



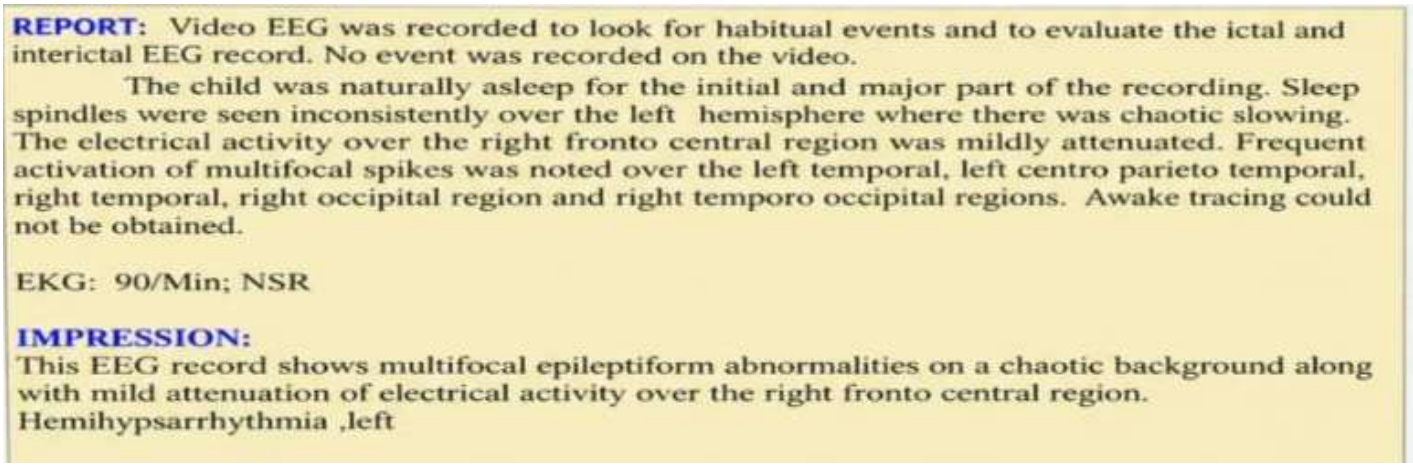
IMPRESSION:

- Open lip schizencephaly in right parietal region.
- Multiple small nodular calcifications in bilateral frontal and parietal periventricular regions.

  - Possibility of intra uterine TORCH infection

Figure 3: impressions of MRI Scan

VEEG Report showed multifocal epileptiform abnormalities on a chaotic background, consistent with severe epileptic encephalopathy. Frequent multifocal spike discharges were observed involving the left temporal, left centro-parietotemporal, right temporal, right occipital, and right temporo-occipital regions. In addition, there was mild attenuation of electrical activity over the right frontocentral region, suggestive of underlying structural cerebral dysfunction. Sleep spindles were inconsistently seen over the left hemisphere with associated chaotic slowing. The overall EEG pattern was suggestive of left hemihypsarrhythmia, a modified hypsarrhythmic pattern commonly associated with West syndrome. These electrophysiological findings correlated with the patient's clinical presentation of epileptic spasms and underlying structural brain abnormalities.



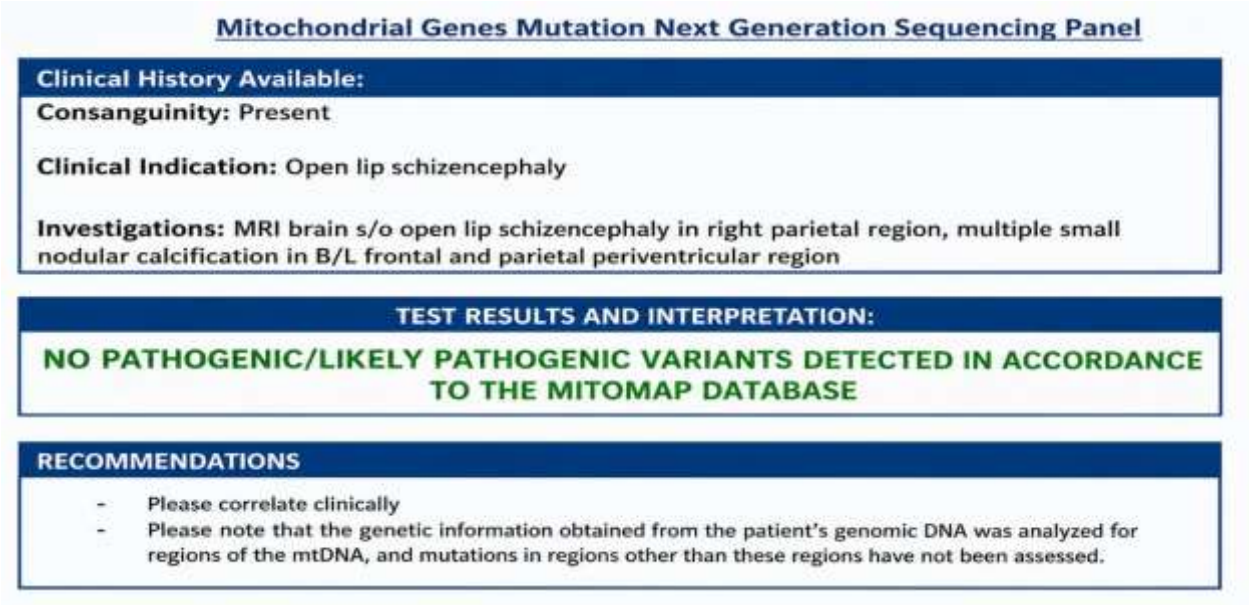
REPORT: Video EEG was recorded to look for habitual events and to evaluate the ictal and interictal EEG record. No event was recorded on the video.

The child was naturally asleep for the initial and major part of the recording. Sleep spindles were seen inconsistently over the left hemisphere where there was chaotic slowing. The electrical activity over the right fronto central region was mildly attenuated. Frequent activation of multifocal spikes was noted over the left temporal, left centro parieto temporal, right temporal, right occipital region and right temporo occipital regions. Awake tracing could not be obtained.

EKG: 90/Min; NSR

IMPRESSION:
This EEG record shows multifocal epileptiform abnormalities on a chaotic background along with mild attenuation of electrical activity over the right fronto central region.
Hemihypsarrhythmia ,left

Figure 4: VEEG Report



Mitochondrial Genes Mutation Next Generation Sequencing Panel

Clinical History Available:

Consanguinity: Present

Clinical Indication: Open lip schizencephaly

Investigations: MRI brain s/o open lip schizencephaly in right parietal region, multiple small nodular calcification in B/L frontal and parietal periventricular region

TEST RESULTS AND INTERPRETATION:

NO PATHOGENIC/LIKELY PATHOGENIC VARIANTS DETECTED IN ACCORDANCE TO THE MITOMAP DATABASE

RECOMMENDATIONS

- Please correlate clinically
- Please note that the genetic information obtained from the patient's genomic DNA was analyzed for regions of the mtDNA, and mutations in regions other than these regions have not been assessed.

Figure 5: Mitochondrial Gene Mutation Next Generation Sequencing (NGS) Panel Report I

The mitochondrial gene mutation analysis was performed using Next Generation Sequencing (NGS) on a whole blood EDTA sample to evaluate possible mitochondrial genetic abnormalities associated with the patient's neurological manifestations. The report was conducted in view of the clinical history of open-lip schizencephaly and developmental neurological abnormalities. Mitochondrial gene analysis helps identify pathogenic or likely pathogenic variants in mitochondrial DNA that may contribute to epileptic encephalopathy, developmental delay, infantile spasms, and neurodevelopmental disorders. The report mentions that phenotype-related mitochondrial

variants were analyzed and interpreted according to the currently available scientific evidence. Such testing is useful in assessing suspected mitochondrial disease and understanding the possible genetic basis of the patient's clinical presentation.

Mitochondrial Gene Mutation Next Generation Sequencing (NGS) Panel analysis was performed to evaluate possible mitochondrial genetic abnormalities in a child with open lip schizencephaly. Clinical history revealed consanguinity, which may increase the risk of inherited genetic disorders. MRI brain findings showed open lip schizencephaly in the right parietal region along with multiple small nodular calcifications in the bilateral frontal and parietal periventricular regions, suggesting abnormal brain development and raising the possibility of intrauterine TORCH infection or other congenital etiologies. The genetic analysis did not detect any pathogenic or likely pathogenic mitochondrial DNA variants according to the MITOMAP database.

These findings indicate that no confirmed mitochondrial gene mutation was identified in the tested mitochondrial DNA regions; however, mitochondrial disease cannot be completely excluded because mutations in untested regions or nuclear genes associated with mitochondrial disorders may not have been assessed. Therefore, the genetic findings should be interpreted in correlation with the clinical presentation, radiological findings, and other laboratory investigations.

The child was treated with NEBULIZATION 3% NORMAL SALINE 3ml P/N BD, INJECTION AMIKACIN 120 mg IV OD, INJECTION CEFOTAXIME 400 mg IV TID, SYP. SODIUM VALPROATE (200mg/5ml) P/O BD, SYP.CLOBAZAM 1ml P/O HS, TAB.PHENOBARBITONE 30mg ½ HS and TAB.LEVOTHYROXINE SODIUM 25 mcg P/O OD, INJECTION PANTOPRAZOLE 40 mg IV OD, INJECTION ONDANSETRON 1 mg IV SOS, NEBULIZATION LEVOSALBUTAMOL+ BUDESONIDE P/N Q8H and SYRUP CEFOTAXIME 4ml P/O BD.

DISCUSSION

West syndrome is a severe epileptic encephalopathy of infancy characterized by the triad of infantile spasms, hypsarrhythmia on electroencephalography, and developmental delay. Structural brain abnormalities are among the most common etiologies, with schizencephaly representing a rare but important cause. In the present case, the coexistence of West syndrome, epileptiform encephalopathy, open-lip schizencephaly, and suspected mitochondrial disease represents an unusual and complex neurodevelopmental association[16].

Schizencephaly is a rare congenital cortical malformation characterized by gray matter-lined clefts extending from the pial surface to the ventricular system. Open-lip schizencephaly is generally associated with more severe neurological manifestations including developmental delay, motor deficits, and refractory seizures. Previous reports have demonstrated that epilepsy is common in patients with schizencephaly; however, the occurrence of West syndrome in association with schizencephaly is rare. Imataka et al[17].. reported a child with type I schizencephaly who developed infantile spasms and hypsarrhythmia, similar to the present case. Their patient showed developmental delay and responded to ACTH therapy, highlighting the epileptogenic potential of cortical malformations in triggering West syndrome.

Similarly, Hapsari et al[18]. described epilepsy associated with open-lip schizencephaly in an adult patient, emphasizing that cortical clefts can serve as persistent epileptogenic foci. Although their patient presented later in life, the report supports the association between open-lip schizencephaly and severe seizure disorders. In the current case, the presence of epileptiform encephalopathy at infancy indicates a more severe neurodevelopmental disruption likely related to extensive cortical malformation.

The coexistence of suspected mitochondrial disease further increases the complexity of this case. Mitochondrial dysfunction has been increasingly recognized as an important cause of epileptic encephalopathy and infantile

spasms. Motobayashi et al[19]. reported an infant with pyruvate dehydrogenase complex deficiency presenting with West syndrome, demonstrating that mitochondrial energy metabolism defects can contribute to refractory epileptic spasms and developmental regression. Likewise, Papetti et al [20]. described severe early-onset ethylmalonic encephalopathy associated with West syndrome, suggesting that mitochondrial abnormalities may play a role in epileptogenesis through impaired neuronal energy production and excitotoxic injury.

In the present case, the simultaneous presence of structural cortical malformation and suspected mitochondrial dysfunction may have synergistically contributed to the severe epileptic phenotype. Structural abnormalities such as schizencephaly disrupt normal cortical organization and neuronal migration, predisposing to abnormal electrical discharges, while mitochondrial dysfunction may further aggravate neuronal hyperexcitability and neurodegeneration. This dual pathology could explain the early onset, epileptiform encephalopathy, and poor neurodevelopmental outcome observed in this infant[21].

Developmental delay and neurological impairment are commonly reported in patients with both West syndrome and schizencephaly. Previous studies have shown that bilateral or open-lip clefts are associated with worse cognitive and motor outcomes compared to closed-lip forms. The presence of epileptic encephalopathy in infancy further worsens prognosis because persistent epileptic activity interferes with brain maturation and synaptic development. Similar findings were observed in previously reported cases of West syndrome secondary to structural brain abnormalities[22].

Management of such patients remains challenging. ACTH, vigabatrin, and other antiepileptic drugs are commonly used in West syndrome; however, treatment response is often limited in symptomatic cases associated with cortical malformations or metabolic disorders. Early diagnosis using neuroimaging and metabolic evaluation is therefore essential for prognostication and multidisciplinary management. In suspected mitochondrial disease, supportive metabolic therapies and genetic evaluation may help in identifying the underlying etiology and guiding counseling for families[23].

Overall, this case highlights a rare coexistence of West syndrome, epileptiform encephalopathy, open-lip schizencephaly, and suspected mitochondrial disease in an infant. The report emphasizes the importance of considering both structural and metabolic etiologies in infants presenting with refractory spasms and developmental delay. Early neuroimaging, EEG evaluation, and metabolic workup are crucial for accurate diagnosis and comprehensive management.

CLINICAL SIGNIFICANCE

The coexistence of West syndrome, epileptiform encephalopathy, open-lip schizencephaly, and suspected mitochondrial disease in an infant is extremely rare and clinically significant. This case highlights the importance of considering both structural and metabolic causes in infants presenting with refractory seizures, developmental delay, and abnormal neurological findings. Open-lip schizencephaly, a severe cortical malformation, can act as a structural substrate for epileptogenesis, while suspected mitochondrial dysfunction may further aggravate neuronal injury and seizure activity through impaired cellular energy metabolism.

The case emphasizes the value of early neuroimaging, electroencephalography, and metabolic investigations in identifying the underlying etiology of infantile spasms and epileptic encephalopathy. Early diagnosis is crucial because persistent epileptic activity during infancy can interfere with brain maturation, resulting in long-term cognitive and motor impairment. Recognition of possible mitochondrial involvement is also clinically important for appropriate drug selection and prevention of metabolic complications.

In addition, this rare association contributes to the limited literature on complex neurodevelopmental disorders and may improve understanding of the pathophysiological relationship between cortical malformations, mitochondrial dysfunction, and severe epilepsy syndromes.

THERAPEUTIC CHALLENGES

Management of this condition is challenging because of the coexistence of severe epileptic encephalopathy, structural brain abnormality, and suspected metabolic dysfunction. Seizures associated with symptomatic West syndrome are often refractory to conventional treatment, and therapeutic response to ACTH, corticosteroids, or vigabatrin may be limited. The suspicion of mitochondrial disease further complicates treatment, as certain antiepileptic drugs such as valproate may worsen mitochondrial dysfunction and increase the risk of metabolic toxicity. Careful selection of medications and close monitoring are therefore necessary.

The patient may also experience significant neurodevelopmental impairment including developmental delay, motor dysfunction, cognitive deficits, and feeding difficulties, requiring long-term multidisciplinary rehabilitation and supportive care. Additionally, confirmation of mitochondrial disease often requires advanced metabolic and genetic investigations, which may not always be accessible in all healthcare settings. Overall, the coexistence of multiple neurological abnormalities contributes to poor prognosis, increased treatment complexity, and the need for individualized multidisciplinary management to optimize seizure control and improve quality of life.

CONCLUSION

This case highlights a rare coexistence of West syndrome, epileptiform encephalopathy, open-lip schizencephaly, and suspected mitochondrial disease in an infant, emphasizing the complex interplay between structural brain malformations and possible metabolic dysfunction in the pathogenesis of severe early-onset epilepsy. The presence of infantile spasms, developmental delay, and refractory epileptiform activity in association with open-lip schizencephaly suggests a poor neurological prognosis and underscores the importance of early neuroimaging and electroencephalographic evaluation in infants presenting with seizures and developmental regression.

The suspected mitochondrial involvement further broadens the etiological spectrum and indicates the need for detailed metabolic and genetic investigations in atypical or severe presentations of epileptic encephalopathy. Early identification of underlying causes may facilitate timely therapeutic interventions, multidisciplinary management, prognostic counseling, and long-term follow-up. This case contributes to the limited literature on the coexistence of these rare neurological conditions and highlights the importance of considering both structural and metabolic etiologies in infants with refractory epilepsy and neurodevelopmental impairment.

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