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Case Report

Mowat-Wilson Syndrome Presenting with Fever-Associated Status Epilepticus: A Case Report

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Abstract

Mowat-Wilson syndrome (MWS) is a rare neurodevelopmental disorder caused by pathogenic variants in the ZEB2 gene. Although epilepsy is common, fever-associated status epilepticus is an uncommon presenting feature.

A 13-month-old girl presented with fever-associated status epilepticus. Neurological examination revealed generalized hypotonia and global developmental delay. Physical examination showed mild facial dysmorphism and unilateral iris coloboma. MWS was suspected and subsequently confirmed by identification of a pathogenic ZEB2 variant.

MWS should be considered in infants presenting with fever-associated status epilepticus, particularly when accompanied by developmental delay, hypotonia, and dysmorphic features. Early molecular diagnosis facilitates appropriate multidisciplinary management and genetic counseling.

Key Words: status epilepticus; developmental delay; coloboma

Introduction

Mowat-Wilson syndrome (MWS) is a rare autosomal dominant neurodevelopmental disorder caused by heterozygous pathogenic variants in the *ZEB2* gene. It is characterized by developmental delay, intellectual disability, distinctive facial features, epilepsy, and variable congenital anomalies [1-4]. Epilepsy is a common feature of MWS, with seizure onset typically occurring during infancy or early childhood. Although febrile seizures are frequently reported, fever-associated status epilepticus is an uncommon presenting manifestation [2-4].

Here, we report a genetically confirmed case of MW presenting with fever-associated status epilepticus. This case highlights the importance of recognizing the characteristic clinical features of MWS in infants presenting with prolonged febrile seizures to facilitate early diagnosis, appropriate management, and genetic counseling.

Case Presentation

A 13-month-old girl was admitted to the pediatric emergency department with fever-associated status epilepticus. She had no previous history of afebrile seizures. Status epilepticus was managed according to the institutional treatment protocol, resulting in seizure cessation. Maintenance therapy with levetiracetam was subsequently initiated, and no further seizures occurred during hospitalization.

She was born by cesarean section at 37 weeks of gestation with a birth weight of 2850 g following an uncomplicated pregnancy and delivery. Neurological examination revealed generalized hypotonia with preserved deep tendon reflexes. Developmental assessment demonstrated global developmental delay. She had achieved head control later than expected and was able to sit only with support but was unable to sit independently at 13 months of age.

Physical examination revealed mild facial dysmorphism and unilateral iris coloboma. The combination of global developmental delay, hypotonia, dysmorphic facial features, and iris coloboma raised suspicion of an underlying genetic syndrome. Brain magnetic resonance imaging, abdominal ultrasonography and echocardiography revealed no abnormalities. EEG was normal. Molecular genetic testing identified a pathogenic *ZEB2* variant, confirming the diagnosis of MWS.

Discussion

Developmental delay and hypotonia are among the earliest neurological manifestations of MWS. Characteristic facial features become more evident with age and, together with associated congenital anomalies, provide important diagnostic clues. Ocular abnormalities, including iris coloboma, although uncommon, have also been reported [3,4].

Epilepsy is a common manifestation of MWS, affecting approximately 75-80% of patients during childhood [5]. Seizures usually begin in early childhood, often triggered by fever, and include febrile, focal, generalized tonic-clonic seizures, and less commonly, status epilepticus. Experimental studies suggest that ZEB2 plays a key role in the migration and differentiation of GABAergic interneurons, and pathogenic variants may disrupt the balance between excitatory and inhibitory neuronal

networks, predisposing patients to epilepsy [6]. Seo et al. reported a 4-year-old girl with recurrent fever-associated generalized tonic-clonic seizures beginning at 10 months of age [1]. Bonanni et al. described an 11-year-old boy with electrical status epilepticus during sleep (ESES) associated with language regression and cognitive decline [7]. Ju et al. reported five children with seizure onset between 8 months and 6 years, in whom febrile and afebrile seizures were the most common initial manifestations [8]. Paz et al. reported that most of the seven patients with MWS had epilepsy, characterized by early childhood onset, variable seizure types, and frequent EEG abnormalities [3]. In contrast, our patient presented with fever-triggered status epilepticus as the initial manifestation and, to the best of our knowledge, represents the first reported case of MWS presenting in this manner.

The combination of fever-associated status epilepticus, global developmental delay, hypotonia, mild facial dysmorphism, and iris coloboma raised suspicion of an underlying genetic syndrome, which was confirmed by identification of a pathogenic *ZEB2* variant. This case emphasizes the importance of considering MWS in infants with prolonged fever-associated seizures or status epilepticus, particularly when developmental delay and characteristic dysmorphic features are present.

In conclusion, fever-triggered status epilepticus may represent an uncommon initial manifestation of MWS. Early recognition and molecular diagnosis are essential for appropriate multidisciplinary management, surveillance for associated anomalies, and genetic counseling.

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