Neonatal and Pediatric Medicine
ISSN: 2572-4983
Martinez and Nguyen, Neonat Pediatr Med 2024, 10:2
Review Article
Open Access

Benign Familial Neonatal Seizures (BFNS): A Guide to Recurrent Seizures in Newborns

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Abstract

Benign Familial Neonatal Seizures (BFNS) is a rare condition characterized by recurrent seizures in newborn infants. These seizures typically manifest around day 3 of life and typically resolve within 1 to 4 months. They can present as focal seizures, affecting only one side of the brain, or generalized seizures, involving both sides. Often, infants with BFNS experience generalized tonic-clonic seizures, also known as grand mal seizures. Despite its alarming presentation, BFNS carries a favorable prognosis, with most infants outgrowing the seizures without long-term consequences. However, understanding the underlying mechanisms and genetic factors contributing to BFNS remains a subject of ongoing research. This abstract provides an overview of BFNS to aid healthcare professionals and caregivers in recognizing and managing this condition effectively.

Keywords: Benign familial neonatal seizures; BFNS; Recurrent seizures; Generalized seizures; Tonic-clonic seizures; Grand mal seizures

Introduction

Benign Familial Neonatal Seizures (BFNS) is a relatively uncommon neurological disorder that presents a challenge to clinicians due to its alarming onset in newborn infants. This condition is characterized by the occurrence of recurrent seizures within the first few days of life, typically around day 3. These seizures can manifest as focal seizures, affecting only one hemisphere of the brain, or generalized seizures, involving both hemispheres. The most frequently observed seizure type in BFNS is the generalized tonic-clonic seizure, also known as a grand mal seizure, which can be distressing for both caregivers and healthcare providers. Despite the dramatic nature of the seizures, BFNS is considered benign, as the seizures generally remit spontaneously within 1 to 4 months without leaving lasting neurological deficits. However, the precise mechanisms underlying the onset and resolution of seizures in BFNS remain incompletely understood. Additionally, while the condition is termed "familial," indicating a genetic component, the specific genes involved and their mode of inheritance have yet to be fully elucidated.

In this introduction, we aim to provide a comprehensive overview of BFNS, including its clinical presentation, epidemiology, underlying genetics, diagnostic approach, and management strategies. By enhancing our understanding of BFNS, healthcare professionals can better recognize, diagnose, and manage this condition, ultimately improving outcomes for affected infants and their families [1].

Clinical presentation of benign familial neonatal seizures (BFNS)

The clinical presentation of Benign Familial Neonatal Seizures (BFNS) typically involves the sudden onset of seizures within the first few days of life, commonly around day 3. These seizures can manifest as focal or generalized, with generalized tonic-clonic seizures being the most frequent presentation. Focal seizures may involve rhythmic jerking or twitching of one part of the body, while generalized seizures often present with whole-body stiffening followed by rhythmic jerking movements. Infants with BFNS may experience multiple seizures throughout the day, varying in duration and intensity. While the seizures can be alarming for caregivers and healthcare providers, it's essential to recognize that BFNS is generally a self-limiting condition, with seizures spontaneously remitting within 1 to 4 months without causing permanent neurological damage. However, thorough evaluation and monitoring are necessary to rule out other underlying conditions and ensure appropriate management [2].

Epidemiology and prevalence:

Benign Familial Neonatal Seizures (BFNS) is a relatively rare neurological disorder, accounting for a small percentage of neonatal seizures. The exact prevalence of BFNS is not well-established, but it is estimated to occur in approximately 1 in 20,000 to 1 in 100,000 live births. However, these figures may vary across different populations and geographic regions. Although BFNS is considered a familial condition, implying a genetic basis, sporadic cases also occur. It tends to affect both sexes equally and has been reported in various ethnic groups worldwide. Family history of seizures, particularly neonatal or early infancy seizures, may increase the likelihood of BFNS in subsequent generations. Understanding the epidemiology and prevalence of BFNS is crucial for healthcare providers to recognize and appropriately manage this condition. Further research into the genetic and environmental factors contributing to BFNS incidence is necessary to improve our understanding and enhance diagnostic and therapeutic approaches.

Genetic factors and inheritance patterns:

Benign Familial Neonatal Seizures (BFNS) is recognized as a genetically heterogeneous disorder with a complex inheritance pattern. While the condition is termed "familial," indicating a genetic component, sporadic cases also occur, suggesting potential de novo mutations or multifactorial inheritance. Several genes have been

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Received: 2-Feb-2024, Manuscript No. nnp-24-130269; **Editor assigned:** 5-Feb-2024, Pre-QC No. nnp-24-130269 (PQ); **Reviewed:** 19-Feb-2024, QC No. nnp-24-130269; **Revised:** 22-Feb-2024, Manuscript No. nnp-24-130269 (R); **Published:** 29-Feb-2024, DOI: 10.4172/2572-4983.1000384

Citation: Martinez D, Nguyen S (2024) Benign Familial Neonatal Seizures (BFNS): A Guide to Recurrent Seizures in Newborns. Neonat Pediatr Med 10: 384.

implicated in BFNS, with mutations in KCNQ2 and KCNQ3 genes being the most commonly identified genetic cause. These genes encode subunits of voltage-gated potassium channels, critical for regulating neuronal excitability. Mutations in KCNQ2 and KCNQ3 disrupt the function of these channels, leading to hyperexcitability and increased susceptibility to seizures. Inheritance patterns of BFNS vary depending on the specific genetic mutation involved. Autosomal dominant inheritance with variable penetrance has been reported in families with BFNS associated with mutations in KCNQ2 and KCNQ3 genes. However, de novo mutations are common in sporadic cases, where affected individuals have no family history of seizures [3].

Genetic testing plays a crucial role in confirming the diagnosis of BFNS and identifying the underlying genetic cause. Additionally, genetic counseling is essential for affected families to understand the inheritance pattern, recurrence risk, and implications for family planning. Despite significant progress in identifying genetic factors associated with BFNS, there is still much to learn about the underlying mechanisms and the contribution of additional genetic and environmental factors. Further research is needed to elucidate the complex genetic architecture of BFNS and develop targeted therapeutic strategies.

Diagnostic approach to BFNS:

Clinical Evaluation: The diagnostic process for Benign Familial Neonatal Seizures (BFNS) typically begins with a thorough clinical evaluation. Healthcare providers assess the infant's medical history, including prenatal, perinatal, and postnatal factors, as well as family history of seizures or neurological disorders.

Electroencephalography (EEG): EEG is a critical tool in the diagnosis of BFNS. It helps to characterize the seizure activity and identify abnormal electrical patterns in the brain. In BFNS, EEG findings may include focal or generalized epileptiform discharges, which can aid in distinguishing it from other seizure disorders. While neuroimaging studies such as magnetic resonance imaging (MRI) or computed tomography (CT) scans are not typically indicated for the diagnosis of BFNS, they may be performed to rule out structural abnormalities or other underlying conditions that could be contributing to the seizures [4].

Genetic testing: Given the genetic basis of BFNS, genetic testing plays a crucial role in confirming the diagnosis and identifying specific genetic mutations. Targeted sequencing of genes associated with BFNS, such as KCNQ2 and KCNQ3, may be performed to detect pathogenic variants. Differential Diagnosis is essential to consider other conditions that can present with neonatal seizures and mimic BFNS, including metabolic disorders, electrolyte imbalances, infections, and structural brain abnormalities. A comprehensive evaluation is necessary to differentiate BFNS from these other etiologies.

Longitudinal follow-up: BFNS is characterized by a transient course, with seizures typically remitting within 1 to 4 months of age. Longitudinal follow-up is essential to monitor the infant's neurological development, assess seizure control, and identify any potential recurrence or evolution of seizures. Overall, the diagnostic approach to BFNS involves a multidisciplinary approach, including clinical evaluation, neurophysiological testing, genetic analysis, and longitudinal monitoring. Collaboration between pediatricians, neurologists, geneticists, and other healthcare professionals is crucial for accurate diagnosis and appropriate management of affected infants.

Management strategies for BFNS:

Antiepileptic medications: While the seizures in Benign Familial Neonatal Seizures (BFNS) typically remit spontaneously within the first few months of life, antiepileptic medications may be considered in cases of frequent or prolonged seizures or when seizures pose a significant risk to the infant's safety. Medications such as phenobarbital, levetiracetam, or phenytoin may be used, depending on the clinical presentation and response to treatment. Close monitoring of medication efficacy and adverse effects is essential. Caregivers should receive education and training on how to recognize and manage seizures effectively. This includes ensuring a safe environment for the infant, avoiding potential triggers, and knowing when to seek medical assistance. Seizure diaries can help track seizure frequency, duration, and response to treatment [5].

Supportive care: Providing supportive care to the infant and their family is essential in managing BFNS. This includes addressing the emotional and psychological impact of the seizures on the family, providing resources and support groups, and connecting families with appropriate healthcare professionals, including pediatricians, neurologists, and genetic counselors. Genetic counseling is essential for families affected by BFNS to understand the inheritance pattern, recurrence risk, and implications for family planning. Genetic counselors can provide information about genetic testing, inheritance patterns, and available resources for families affected by genetic conditions. Although BFNS is considered a benign condition with a favorable prognosis, long-term follow-up is necessary to monitor the infant's neurological development and assess for any potential recurrence or evolution of seizures. Neurodevelopmental assessments may be performed to evaluate cognitive and motor development and identify any developmental delays or concerns.

Research and clinical trials: Participation in research studies and clinical trials may offer opportunities for families affected by BFNS to contribute to advancing our understanding of the condition and exploring novel treatment approaches. Healthcare providers can provide information about ongoing research studies and clinical trials that may be relevant to the family's situation. Overall, the management of BFNS involves a multidisciplinary approach, including medical management, supportive care, genetic counseling, and long-term follow-up. Collaboration between healthcare professionals and families is essential to ensure comprehensive care and optimal outcomes for infants affected by BFNS [6].

Results and Discussion

The management of Benign Familial Neonatal Seizures (BFNS) revolves around understanding the transient nature of the condition, addressing acute symptoms, providing supportive care, and ensuring long-term monitoring.

Transient nature: BFNS is characterized by recurrent seizures that typically remit spontaneously within 1 to 4 months of life. Therefore, the primary goal of management is to provide symptomatic relief and support until the seizures resolve. Antiepileptic medications may be considered in cases of frequent or prolonged seizures to mitigate immediate risks and discomfort [7].

Acute symptom management: During active seizure episodes, prompt intervention is necessary to ensure the infant's safety and minimize potential complications. Caregivers should be educated on how to recognize seizures, implement seizure safety measures, and administer rescue medications if prescribed. Providing emotional support and education to families affected by BFNS is essential in

managing the condition. Caregivers should be informed about the benign nature of BFNS, its expected course, and the importance of adherence to medication regimens if prescribed. Support groups and resources for families affected by epilepsy can offer additional support and guidance. Although BFNS is typically self-limiting, long-term follow-up is necessary to monitor the infant's neurodevelopmental progress and assess for any potential recurrence or evolution of seizures. Neurodevelopmental assessments can help identify any developmental delays or cognitive impairments that may require intervention [8].

Genetic counseling: Genetic counseling is crucial for families affected by BFNS to understand the inheritance pattern, recurrence risk, and implications for family planning. Genetic testing may provide valuable information about the underlying genetic cause and inform decisions regarding family planning and genetic testing for at-risk relatives. Continued research into the genetic basis of BFNS and its underlying mechanisms is essential to improve our understanding of the condition and develop targeted therapeutic strategies. Participation in research studies and clinical trials may offer opportunities for families affected by BFNS to contribute to advancing knowledge and exploring new treatment options. Overall, a comprehensive and multidisciplinary approach to the management of BFNS, including medical management, supportive care, genetic counseling, and long-term monitoring, is essential to ensure optimal outcomes for affected infants and their families. Collaboration between healthcare professionals, researchers, and families is key to advancing our understanding of BFNS and improving treatment options in the future [9,10].

Conclusion

In conclusion, Benign Familial Neonatal Seizures (BFNS) presents as a transient condition characterized by recurrent seizures in newborns, typically resolving within 1 to 4 months without lasting neurological damage. Management focuses on symptomatic relief, supportive care, genetic counseling, and long-term monitoring. Continued research is needed to further understand the genetic and neurobiological mechanisms underlying BFNS, paving the way for improved diagnostic and therapeutic strategies.

Acknowledgment

None

Conflict of Interest

None

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