Review article

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Caring for Children with ARPC1B Deficiency Syndrome

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ABSTRACT

ARPC1B Deficiency Syndrome is a rare genetic disorder recently characterized as an autosomal recessive condition linked to mutations in the ARPC1B gene. This gene encodes a subunit of the actin-related protein 2/3 (Arp2/3) complex, impacting the actin cytoskeleton and resulting in a variety of symptoms and severities. The syndrome primarily presents as a combined immune deficiency, leading to recurrent infections, allergic reactions, and inflammatory conditions. Due to its rarity and recent discovery, the exact mechanism and full spectrum of severity remain unknown. This paper aims to provide comprehensive nursing management for children with ARPC1B Deficiency Syndrome, focusing on assessment, monitoring, intervention, and support to address physical, developmental, and psychosocial needs. The approach includes monitoring growth and developmental milestones, immune function, and emotional well-being, providing education on infection prevention, developmental therapies, and genetic counseling to families. This strategy is to improve health outcomes and enhance quality of life by offering ongoing support and education.

Keywords: *ARPC1B deficiency, ARPC1B gene, ARP2/3 complex, combined immune deficiency, Nursing care*

INTRODUCTION

ARPC1B Deficiency Syndrome is a rare genetic disorder affecting the immune system and other physiological functions. It is characterized by combined immune deficiency, recurrent infections, allergies, asthma, and auto inflammation, with an increased risk of severe infections. The syndrome is also known as Platelet Abnormalities with Eosinophilia and Immune-Mediated Inflammatory Disease (PTLEID) or Immunodeficiency-71 with Inflammatory Disease and Congenital Thrombocytopenia (IMD71).[1] This globally rare condition has limited understanding due to its recent discovery and rarity, affecting both sexes equally.[2] Earlier case studies revealed multiple disease-causing variants in the ARPC1B gene, leading to diverse symptoms such as recurrent infections, eczema, food allergies, asthma, and thrombocytopenia.[3]

There are currently no specific reports of ARPC1B deficiency syndrome cases in Nepal. However, a recent study highlighted the molecular profile of patients with inborn errors of immunity in Nepal, marking the first report of genetically proven immunodeficiencies in the country[4]. Recently, an 8-year-old female child referred from Kanti Children's Hospital was admitted to the Pediatric Department of Tribhuvan University Teaching Hospital. Therefore, we are interested in writing this paper.

Causes

ARPC1B Deficiency is an autosomal recessive disease resulting from genetic mutations in the ARPC1B gene. This gene encodes a subunit of the actin-related protein 2/3 (ARP2/3) complex, crucial for regulating the actin cytoskeleton in cells.[5] Consanguineous marriages and a family history of ARPC1B deficiency are considered risk factors for this condition.[2]

Clinical manifestations

Clinical features of ARPC1B deficiency vary but commonly include immune system abnormalities especially combined immune deficiency, developmental issues, and systemic manifestations like severe lower respiratory tract infection (LRTI) with wheezing and difficulty in breathing. Children with ARPC1B deficiency often experience severe infections, developmental delays eg hypothyridism, cognitive impairment, growth retardation, dry skin, scaly skin. Chronic dermatitis, allergies, asthma, and thrombocytopenia.[2,3,5,6] Some

may also present with seizures and other neurological conditions.[2]

Diagnostic evaluation

Diagnosing ARPC1B deficiency involves clinical evaluation, laboratory tests (total leucocyte count, differential count, CD4 and CD8 marker, IgG, IgM level, and genetic analysis to confirm mutations in the ARPC1B gene.[1,2] Genetic sequencing is the definitive diagnostic test, with radiological studies used to assess complications from infections. Genetic panels may be employed to identify related disorders with similar symptoms.[2,3,5-7]

Treatment and prognosis

There is no definitive treatment for ARPC1B deficiency, requiring a multidisciplinary approach tailored to the child's needs. Regular monitoring, prompt infection treatment, and vaccines for prevention are essential.[7] Apart form infection control, preventive therapy for asthma and other conditions, Physical, occupational, and speech therapies can address developmental delays and neurological symptoms.[8] Genetic counseling for parents and caregivers is also important.[9] Gene therapy approaches targeting the underlying genetic mutation hold promise but are still experimental.[10] Hematopoietic Stem Cell Transplantation (HSCT) is an option for severe cases with profound immunodeficiency.[11]

The prognosis for a child with APRC1B deficiency syndrome varies depending on the severity of symptoms and the effectiveness of treatment. Early diagnosis and prompt management of infections, allergies, and autoimmune manifestations are crucial to improve outcomes.[2,3,5]

Nursing care of a child with arpc1b deficiency syndrome

Navigating ARPC1B Deficiency Syndrome requires a comprehensive holistic approach to managing a child with ARPC1B deficiency. The nursing management of this syndrome involves a multifaceted approach that includes monitoring, intervention, and support to meet physical, developmental, and psychosocial needs, as well as ongoing evaluation of the child. The following structured and detailed guide provides a nursing perspective on navigating ARPC1B Deficiency Syndrome.[12-17]

Assessment and Monitoring

1. Regularly monitor growth and development milestones to identify delays or abnormalities.
2. Assess for signs of muscle weakness, skeletal deformities, and potential complications
3. Monitor immune function and watch for signs of frequent infections or other health problems.
4. Evaluate the child's and family's emotional well-being.
5. Assess the family's understanding of ARPC1B deficiency and its impact.

Diagnosis

1. Impaired physical mobility related to muscle weakness or skeletal abnormalities.
2. Risk for infection related to potential immune system compromise.
3. Delayed growth and development related to the overall impact of the condition on physical and cognitive development.
4. Knowledge deficit related to the family's understanding of the condition and its management.

Planning and Interventions

Impaired physical mobility

1. Collaborate with physical therapists to develop and implement an individualized exercise and therapy program to improve mobility and strength
2. Encourage engagement in age-appropriate activities
3. Monitor for and manage any signs of discomfort or pain and other complications.

Risk for infection

1. Educate the family on infection prevention strategies, including proper hand hygiene and recognizing early signs of infection.
2. Ensure the child is up-to-date with vaccinations and follow any prophylactic treatments as recommended.
3. Delayed growth and development
4. Coordinate with developmental specialists (e.g., occupational therapists, speech therapists) to provide targeted therapies.
5. Support developmental milestones through structured activities and therapies

Family support and education

Managing a child with ARPC1B deficiency requires a lot of family education, counseling, and support. For affected children and their families, this uncommon genetic disorder can pose serious difficulties Providing comprehensive, clear, and empathetic support can help families navigate the complexities of the condition, make informed decisions, and access appropriate resources.

1. Discuss common symptoms of ARPC1B deficiency, including developmental delays, immune system issues, muscle weakness, and skeletal abnormalities
2. Provide information on potential outcomes and progression of the disease
3. Educate families about management strategies and resources, including regular medical follow-ups and therapies
4. Explain the genetic basis of the condition and provide strategies for infection prevention
5. Encourage participation in support groups and self-care for caregivers
6. Support families in making informed decisions about their child's care and communicating with healthcare providers.

Evaluation and Follow-up

1. Assess progress in physical therapy and make adjustments as needed based on periodic evaluations
2. Monitor for any signs of infection and review the effectiveness of infection prevention measures
3. Regularly assess developmental milestones and modify therapeutic interventions based on progress
4. Evaluate the family's understanding of the condition and their ability to manage care effectively, providing additional support or education as required.

CONCLUSIONS

ARPC1B Deficiency Syndrome is a rare genetic disorder that causes combined immune deficiency syndrome. It requires a comprehensive and multidisciplinary nursing approach to effectively manage its diverse symptoms and complications. Nursing care for children with combined immune deficiency syndrome focuses on infection prevention, education, nutritional support, coordination of care, and treatment administration. The primary conclusion is that continuous assessment, monitoring, customized nursing interventions, and strong family support are crucial for enhancing the quality of life for affected children and their families.

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