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Perspective

Managing Fragile X Syndrome in Neonates: A Multidisciplinary Approach for Support and Care

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DESCRIPTION

Fragile X syndrome is a genetic disorder that affects neonates and has significant implications for their development and well-being. It is one of the most common inherited forms of intellectual disability and is caused by a mutation in the *FMR1* gene located on the X chromosome. To explore Fragile X syndrome in neonates, including its causes, symptoms, diagnosis, and potential implications for early intervention and management.

Fragile X syndrome is caused by a mutation in the *FMR1* gene, which leads to a lack or reduced production of a protein called Fragile X Mental Retardation Protein (FMRP). This protein is essential for normal brain development and function, and its absence or deficiency results in the characteristic features of Fragile X syndrome.

The mutation in the *FMR1* gene is a triplet repeat expansion, specifically a CGG repeat sequence. Individuals with Fragile X syndrome have an excessive number of CGG repeats, typically more than 200 repeats, whereas the normal range is around 5 to 55 repeats. The number of CGG repeats is associated with the severity of the syndrome, with a larger number of repeats generally resulting in more significant cognitive and behavioural impairments.

Fragile X syndrome is characterized by a wide range of symptoms and clinical features that can vary in severity among affected individuals. In neonates, the signs of Fragile X syndrome may not be apparent immediately, but early indicators can be observed as the child grows.

Some common symptoms and features of Fragile X syndrome in neonates include

Developmental delays: Neonates with Fragile X syndrome may exhibit delays in reaching developmental milestones, such as sitting up, crawling, or walking. These delays may become more noticeable as the child grows older.

Intellectual disability: Fragile X syndrome is associated with varying degrees of intellectual disability, ranging from mild to severe. It can affect cognitive functioning, learning abilities, and language development.

Speech and language difficulties: Many neonates with Fragile X syndrome experience speech and language delays. They may have trouble with articulation, vocabulary acquisition, and understanding complex language structures.

Behavioral challenges: Behavioral issues are common in Fragile X syndrome, including hyperactivity, attention deficits, impulsivity, social anxiety, and repetitive behaviors. These challenges may impact the child's ability to interact with others and participate in social activities.

Physical features: Some neonates with Fragile X syndrome may exhibit physical characteristics such as a long face, large ears, soft skin, and flexible joints. However, these features can be subtle and may not be apparent until later in childhood.

Diagnosing Fragile X syndrome in neonates typically involves genetic testing to identify the presence of the *FMR1* gene mutation. This can be done through a blood sample or cheek swab to analyses the number of CGG repeats in the gene.

Early diagnosis is crucial for neonates with Fragile X syndrome, as it allows for early intervention and management strategies to be implemented. Early intervention programs may include speech and language therapy, occupational therapy, behavioral interventions, and educational support tailored to the specific needs of the child. These interventions aim to optimize the child's development, address any challenges or delays, and enhance their overall quality of life.

Managing Fragile X syndrome in neonates requires a multidisciplinary approach involving healthcare professionals, therapists, educators, and support networks. Regular medical check-ups and monitoring of developmental progress are essential to address any emerging needs or challenges. Genetic counselling can also be beneficial for families, providing

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information about the condition, recurrence risks, and family planning options.

In conclusion, Fragile X syndrome is a genetic disorder that can significantly impact neonates and their development. The absence or deficiency of the fragile X mental retardation protein due to a mutation in the *FMR1* gene leads to various symptoms and clinical features, including developmental delays, intellectual disability, speech and language difficulties, behavioral challenges, and physical characteristics. Early diagnosis through genetic testing is crucial to initiate early intervention programs and support *tailored* to the child's specific needs. A multidisciplinary approach involving healthcare professionals, therapists, educators, and support networks is essential for managing Fragile X syndrome in neonates and providing the necessary resources and support to families. By understanding the causes, symptoms, and implications of Fragile X syndrome in neonates, they can strive to improve their well-being and enhance their quality of life.