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Short Communication

Down Syndrome: Genetic Translocations and Risk Factors

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DESCRIPTION

Down syndrome (DS) is a chromosomal disorder that occurs when a person has an extra copy of chromosome 21. It is the most common genetic cause of intellectual disabilities, affecting approximately 1 in every 700 births worldwide. While the exact cause of Down syndrome remains unknown, several risk factors have been identified. Understanding these risk factors is crucial for early detection, diagnosis and proper management of the condition [1].

Risk factors

Advanced maternal age: One of the most well-established risk factors for Down syndrome is advanced maternal age. The incidence of Down syndrome increases as a woman gets older. While the risk is present at any age, it significantly rises after the age of 35. The reason behind this association is not fully understood, but it is believed to be related to the aging process of eggs in the ovaries. As women age, there is an increased likelihood of chromosomal errors during meiosis, leading to the formation of eggs with an extra copy of chromosome 21 [2,3].

Prior history of down syndrome: Couples who have previously had a child with Down syndrome are at a higher risk of having another child with the condition. The risk varies depending on the type of Down syndrome present in the previous child. For instance, if the previous child had the non-inherited form of Down syndrome (trisomy 21), the risk of recurrence is low. However, if the child had the inherited form (translocation Down syndrome), the risk of recurrence is higher [4,5].

Parental genetic translocation: Having a family history of Down syndrome increases the risk of having a child with the condition. If a parent carries a translocation, which involves rearrangements of genetic material between chromosomes, the chances of having a child with Down syndrome are higher. Translocations can be inherited from a parent or can occur spontaneously during the formation of reproductive cells. Genetic counseling is essential for families with a history of Down syndrome to assess the risk accurately [6].

Prior history of down syndrome: Parents who have previous child with Down syndrome are at an increased risk of having another child with the condition. The risk varies depending on the specific type of Down syndrome the previous child had. If the previous child had the most common form, known as trisomy 21, the recurrence risk is relatively low. However, if the previous child had Down syndrome due to a translocation, the risk of recurrence is higher and warrants further genetic evaluation and counseling [7,8].

Certain genetic variations: Certain genetic variations have been associated with an increased risk of Down syndrome. For instance, individuals with mosaic Down syndrome, where only some cells in the body have an extra copy of chromosome 21, may have a lower risk of physical and cognitive impairments compared to those with full trisomy 21. Other rare genetic variations, such as Robertsonian translocations, can also increase the risk of having a child with Down syndrome [9].

Environmental factors: While the primary cause of Down syndrome is genetic, some environmental factors have been suggested to play a role. However, the evidence linking environmental factors to Down syndrome is limited and inconclusive. Maternal exposure to certain chemicals or toxins during pregnancy, such as radiation or certain medications, has been hypothesized to increase the risk [10,11].

CONCLUSION

Down syndrome is a complex condition with various risk factors. Recognizing these risk factors is crucial for early detection and diagnosis of Down syndrome, allowing for appropriate medical care and support to be provided to individuals and families affected by the condition. Genetic counseling and prenatal testing can help families assess their individual risk and make informed decisions. On-going studies in the field of genetics and prenatal screening will continue to shed light on the risk factors associated with Down syndrome.

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