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Opinion Article

The Role of Genetics in Childhood Joint Hypermobility and Treatment

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ABOUT THE STUDY

Joint hypermobility in childhood refers to a condition in which a child's joints have an increased range of motion beyond what is considered normal for their age. While hypermobility can be a benign feature, in some cases, it may be associated with various symptoms and disorders. Understanding its etiology, implications, and treatment options is essential for managing the condition effectively.

Etiology of joint hypermobility

Joint hypermobility occurs when the connective tissues, particularly ligaments, tend to be more flexible than usual. This increased flexibility allows the joints to move beyond the normal range of motion. There are several causes and contributing factors to joint hypermobility:

Genetic factors: A primary cause of joint hypermobility in children is genetic. In many cases, it runs in families, suggesting an inherited predisposition. The condition is often linked to the genes that affect the collagen structure and function. Collagen is the protein that provides structure and strength to connective tissues like tendons and ligaments. Mutations or alterations in collagen genes can result in more elastic ligaments, which lead to hypermobility.

Joint laxity: In some children, the ligaments may be more elastic or less stiff than usual, resulting in laxity. This condition is typically present from birth and can affect multiple joints. It may also be associated with a lack of muscle tone, which further exacerbates the hypermobility.

Connective tissue disorders: In rare cases, hypermobility can be associated with underlying connective tissue disorders, such as Ehlers-Danlos Syndrome (EDS) or Marfan syndrome. These conditions often have other systemic implications, including skin elasticity, blood vessel fragility, and cardiovascular issues.

Environmental factors: While genetics plays a major role, environmental factors such as physical activity and posture can contribute to joint hypermobility. Children who engage in activities that require significant flexibility, such as gymnastics or dance, may develop hypermobility over time, although this is usually temporary and not as severe as genetic hypermobility.

Symptoms and diagnosis

Joint hypermobility in children may or may not cause noticeable symptoms. In many cases, it is asymptomatic, with children showing no pain or limitations in their activities. However, in some instances, hypermobility can lead to:

Joint pain: Children with hypermobile joints may experience intermittent joint pain, particularly after physical activities or prolonged use of the affected joints.

Increased risk of injuries: Hypermobile joints are more prone to sprains, strains, and dislocations, especially in active children who engage in sports.

Fatigue and muscle weakness: Children may experience muscle fatigue due to the extra effort required to stabilize the hypermobile joints.

Diagnosing joint hypermobility typically involves a clinical assessment. The beighton Score, a 9-point scale used to assess hypermobility, is commonly used in children. A score of 4 or higher is generally indicative of joint hypermobility.

Treatment of joint hypermobility

In most cases, joint hypermobility in children does not require treatment. Many children outgrow the condition as they age, with a decrease in joint flexibility and an improvement in muscle strength. However, when hypermobility causes pain or functional limitations, treatment is necessary. Treatment options include:

Physical therapy: The primary treatment for children with symptomatic joint hypermobility is physical therapy. A trained physiotherapist can design a customized exercise program to strengthen the muscles surrounding the hypermobile joints, improve stability, and reduce pain. Strengthening exercises help children develop better control over their joints, which can prevent injuries and discomfort.

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Bracing or splinting: In cases where joints are prone to instability or dislocation, braces or splints may be recommended to provide additional support.

Pain management: For children who experience pain, over-the-counter pain relief medications such as acetaminophen or ibuprofen can help manage discomfort. In more severe cases, a doctor may prescribe stronger medications or recommend specific treatments like acupuncture or TENS (Transcutaneous Electrical Nerve Stimulation).

Lifestyle modifications: Children with joint hypermobility may need to make adjustments in their physical activity. Activities that involve high-impact or repetitive joint stress, such as running or jumping, may need to be limited. Low-impact exercises such as swimming or cycling are often recommended to maintain fitness without putting excessive strain on the joints.

Monitoring for associated conditions: In some cases, hypermobility may be part of a broader connective tissue disorder. Children with persistent joint hypermobility or additional systemic symptoms should be monitored for conditions like EDS or Marfan syndrome. Genetic counseling and specialized medical care may be necessary in such cases.

Joint hypermobility in childhood is often a benign and self-limiting condition, but when it causes pain or instability, it requires careful management. Understanding its genetic basis, the associated risks, and treatment options is significant for ensuring that children with hypermobile joints can live active and healthy lives. Early intervention with physical therapy and appropriate lifestyle adjustments can help children with joint hypermobility achieve the best outcomes and reduce the risk of long-term complications.