**Perspective**
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# An Overview of Hyperekplexia and Treatment

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## Perspective

Hyperekplexia is a relatively uncommon neurologic condition characterised by hypertonia and severe startle reactions to touch or sonic stimulation. The hypertonia may be mainly truncal, less apparent after a year and attenuated during sleep. Genetic mutations in a number of distinct genes, all of which play a role in glycine neurotransmission, induce classic hyperekplexia. The central nervous system uses glycine as an inhibitory neurotransmitter. Although hyperekplexia is categorized as a hereditary ailment, other diseases can mirror hyperekplexia's heightened startle.

Exaggerated startle reactions to unexpected sensory stimuli and stiffness are the clinical core symptoms of hereditary hyperekplexia (HPX), an inherited neurological condition caused by genetic abnormalities leading to glycinergic inhibitory transmission failure. HPX is an uncommon and underdiagnosed condition that appears soon after birth and improves with age. Establishing the accurate diagnosis early on is critical in order to begin adequate care to relieve stiffness and limit the risk of consequences, such as potentially fatal apnea during stiffness episodes. Hyperekplexia is a word that describes an excessive or exaggerated startle that is not habitual. Hyperekplexia can be a symptom of numerous illnesses, especially when there is pontine pathology; it can also be seen in newborns and toddlers with complicated genetic abnormalities that cause developmental delay and intellectual incapacity, frequently due to an inborn metabolic mistake or brain malformation.

Hereditary hyperekplexia is characterised by increased muscular tone (hypertonia) and an exacerbated startle response to unexpected stimuli, particularly loud noises. Following the startle reaction, newborns become stiff and unable to move for a short amount of time. Some newborns cease breathing during these stiff times, which can be deadly if left untreated. Besides while they are sleeping, babies with hereditary hyperekplexia show hypertonia consistently. Muscle twitches during falling asleep (hypnagogic myoclonus) and movements of the arms or legs while asleep are further indications and symptoms of inherited hyperekplexia. When tapped on the nose, some newborns stretch their heads forward and undergo limb and neck muscular spasms. Recurrent seizures occur in a small percentage of neonates with familial hyperekplexia (epilepsy).

Hereditary hyperekplexia's signs and symptoms usually disappear by the age of one. Older people with genetic hyperekplexia, on the other hand, may still be startled easily and experience bouts of stiffness, which can lead them to fall down. They may continue to suffer hypnagogic myoclonus, or sleep movements. Individuals with this syndrome may develop a limited tolerance for crowded environments and loud noises as they age. People with epilepsy who have genetic hyperekplexia will experience seizures for the rest of their life. Sudden infant death syndrome (SIDS), a leading cause of unexplained mortality in children under the age of one year, may be caused by hereditary hyperekplexia.

The hyperactive startle response and muscular systems are the most common symptoms of HPX and treatments are generally geared at regulating them. There is presently no remedy for the sickness. Anti-anxiety and anti-spastic pharmaceuticals such clonazepam and diazepam, as well as carbamazepine, phenobarbital and other medications, may be employed. Physical or occupational therapy can be used in conjunction with other treatments to help relieve symptoms and assess the need for assistive devices and adaptive equipment (such as strollers, walkers and seating and positioning devices) to improve quality of life. CBT (cognitive behavioral therapy) is another treatment option for anxiety. When a baby has major breathing problems, a breathing or heart rate monitor may be recommended by the doctor to keep a check on the youngster. Parents may also choose to study infant CPR so that they can assist their kid in the event of an emergency [1-5].

## References

1. Mineyko, Aleksandra, Sharon Whiting and Gail E Graham. "Hyperekplexia: treatment of a severe phenotype and review of the literature." Can J Neurol Sci 38 (2011): 411-416.
2. Bode, Anna and Joseph W Lynch. "The impact of human hyperekplexia mutations on glycine receptor structure and function." Mol Brain 7 (2014): 1-12.
3. Thomas, Rhys H, Seo-Kyung Chung, Sian E Wood and Thomas D Cushion, et al. "Genotype-phenotype correlations in hyperekplexia: apnoeas, learning difficulties and speech delay." Brain 136 (2013): 3085-3095.
4. Nigro, Michael A and Hui Cun N Lim. "Hyperekplexia and sudden neonatal death." Pediatr Neurol 8 (1992): 221-225.
5. Stewart, Wendy A, Ellen P Wood, Kevin E Gordon and Peter R Camfield, et al. "Successful treatment of severe infantile hyperekplexia with low-dose clobazam." J Child Neurol 17 (2002): 154-156.

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