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Muscular Dystrophy New Zealand

Central Core Disease

This condition is also known as Central Core Myopathy or Shy-Magee Syndrome, CCD or CCO.

About the condition:

Central core disease is a rare genetic form of congenital myopathy that causes persistent, mild, slow-progressing skeletal muscle weakness around the trunk of the body, such as shoulders, upper arms, upper legs and hips. It affects males and females equally. It can be diagnosed in infancy or later on in life.

Central core disease is characterized by disorganized areas or "cores" in the center of muscle fibers of individuals with the condition, seen under microscope, as well as few mitochondria, and the absence of the sarcoplasmic reticulum.

Babies with the condition can appear floppy, this is called hypotonia, and often have delays in reaching developmental milestones as sitting, standing and walking. There is often also skeletal abnormalities such as abnormal curvature of the spine called scoliosis or kyphosis, hip dislocation, dislocation of the kneecap, clubfoot, either flattening or abnormally high arch of the foot, and joint deformities called contractures that restrict the movement of certain joints such as at the Achilles Tendon site most commonly.

Inheritance:

It is usually inherited in an autosomal dominant way (however there have been some rare cases resulting from autosomal recessive inheritance). It is a result of an alteration in the *RYR1* gene located on the long arm (q) of chromosome 19 (19q13.1) RYR1 provides instructions for making a protein called ryanodine receptor 1 that allows muscles to contract normally. When this protein is altered as a result of the mutation, muscles do not mature normally or contract normally and start to weaken.

Risks:

Some RYR1 genetic alterations increase the risk of malignant hyperthermia. Malignant hyperthermia is a severe and life-threatening reaction to some anesthetic gases and muscle relaxants used for surgeries. A person with Central Core disease is

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able to have surgeries performed using different agents for anesthesia that will not put them at risk of developing malignant hyperthermia.

Diagnosis:

A number of examinations need to be performed to diagnose Central Core Disease, these include: a physical examination to identify muscle weakness, family history, a muscle biopsy that reveals a characteristic appearance of the muscle cells, blood tests to measure levels of enzyme creatine kinase, electromyography (EMG), nerve conduction velocity studies and also genetic testing that identifies a mutation in the *RYR1*.

Treatment:

Treatment focuses on symptom management and individuals with Central Core Disease often benefit from physiotherapy and occupational therapy. Surgery or other orthopedic interventions as braces, in some cases may be necessary to correct any skeletal abnormalities. Other support includes assistance with seating and mobility devices; as well as training for family members on properly handling, exercising, and stretching certain muscles.

It is important to monitor for lung function as breathing may be affected by muscle weakness. Breathing exercises or other breathing support treatments may be beneficial. The use of inhaled salbutamol, was found to significantly increase muscle strength and stamina in six of eight children with Central Core Disease in one study*.

Genetic counseling will be of benefit for affected individuals and their families.

References and resources:

- Genetic and Rare Diseases (GARD) Information Center: https://rarediseases.info.nih.gov/diseases/6014/central-core-disease
- US National Library of Medicine, Genetics Home Reference: https://ghr.nlm.nih.gov/condition/central-core-disease#
- NORD, National Organization for Rare Disorders:

  http://rarediseases.org/rare-diseases/central-core-disease/

*Reference: Messina S, Hartley L, Main M, Kinali M, Jungbluth H, Muntoni F, Mercuri E. Pilot trial of salbutamol in central core and multi-minicore diseases. Neuropediatrics. 2004; 35:262-266.