MODULE PARENTS

ACTIVITY 2. LEARN MORE ABOUT RARE DISEASES

- **TIME:** 10m.
- **RESOURCES:** Text document.
- **LANGUAGE:** ENG / SPA.

DESCRIPTION:

Once you have listened to the stories of the parents with courage, we provide you with detailed information about the symptoms, diagnosis and treatment of the diseases suffered by the protagonists of the videos.

OBJECTIVES:

The main objective of this activity is to increase knowledge about minority diseases and syndromes.

LESSON OVERVIEW:

Rare diseases are rare medical conditions that may be genetic, congenital or acquired, affecting a limited number of people. Although they face significant challenges, there is hope in scientific progress and community support to improve the quality of life of people affected by these diseases. Read the following information carefully and broaden your knowledge about some of these pathologies.

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CONGENITAL MUSCULAR DYSTROPHY

Definition of the disease

Congenital muscular dystrophy (CMD) is a heterogeneous group of neuromuscular disorders with onset at birth or infancy characterized by hypotonia, muscle wasting, weakness or delayed motor milestones. The group includes myopathies with abnormalities at different cellular levels: the extracellular matrix, the dystrophin-associated glycoprotein complex, the endoplasmic reticulum.

Diagnostic methods

Diagnosis is often difficult due to the wide clinical, immunohistochemical and genetic heterogeneity. The diagnosis is based on clinical findings, immunochemical staining on muscle biopsy, and molecular genetic testing. Increased creatine kinase (CK) levels, presence of intellectual deficiency and type of distribution of different signs or symptoms are useful markers to distinguish different forms. CMDs due to collagen VI disorders are complex and may need additional studies in skin fibroblasts.

Management and treatment

Currently there is no curative therapy, but supportive treatments. Besides global and nutritional management, common restrictive respiratory insufficiency may need mechanical respiratory assistance. Orthopedic complications (joint contractures, spinal deformities) are a main concern and preventive or proactive treatment by regular physical and occupational therapies and trunk and limb orthesis is often useful to reduce the severity and course of the deformities.

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X-LINKED NON PROGRESSIVE CEREBELLAR ATAXIA

Definition of the disease

X-linked non progressive cerebellar ataxia is a rare hereditary ataxia characterized by delayed early motor development, severe neonatal hypotonia, non-progressive ataxia and slow eye movements, presenting normal cognitive abilities and absence of pyramidal signs. Frequently patients also manifest intention tremor, mild dysphagia, and dysarthria. Brain MRI reveals global cerebellar atrophy with absence of other malformations or degenerations of the central and peripheral nervous systems.

Symptoms

People with ataxia have problems with coordination and balance. Often people first notice a problem when they realize they have been falling over more than usual, walking in the dark, struggling to walk in a straight line or have become clumsier than you would expect. As the condition progresses walking may become difficult or even impossible, so people may need to use a wheelchair to get about some or all the time.

Management and treatment

Some of the very rare ataxias are treatable (vitamin E and CoQ10 deficiency, gluten ataxia, or episodic ataxias, for example), which makes it so important for people to get a specific diagnosis of the type of ataxia they have if possible. All of those with ataxia can benefit from a multidisciplinary management approach to help them minimize the associated spectrum of complications that may occur and so that they can adapt to life with ataxia and live life to the fullest.

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CONGENITAL DISORDER OF GLYCOSYLATION (CDG SYNDROME)

Definition of the disease

A fast growing group of inborn errors of metabolism characterized by defective activity of enzymes that participate in glycosylation (modification of proteins and other macromolecules by adding and processing of oligosaccharide side chains). This group is comprised of phenotypically diverse disorders affecting multiple systems including the central nervous system, muscle function, immunity, endocrine system, and coagulation. The numerous entities in this group are subdivided, based on the synthetic pathway affected, into disorder of protein N-glycosylation, disorder of protein O-glycosylation, disorder of multiple glycosylation, and disorder of glycosphingolipid and glycosylphosphatidylinositol anchor glycosylation.

Clynical Manifestations

Almost all types of congenital disorders of glycosylation (CDG) present in infancy. Because of the important biologic functions of the oligosaccharides in both glycoproteins and glycolipids, incorrect synthesis of these compounds results in broad multisystem clinical manifestations [Varki 1993] that may include one or more of the following: failure to thrive, developmental delay, hepatopathy, hypotonia/neurologic abnormalities, hypoglycemia, protein-losing enteropathy, eye abnormalities, immunologic findings, skin abnormalities, and skeletal findings [Rymen & Jaeken 2014]. It is becoming increasingly clear that the clinical spectrum can involve individual or multiple organ systems and may or may not affect neurodevelopment. CDG is increasingly being considered in the differential diagnosis for varied symptoms across multiple age groups and clinical specialties.

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CHARCOT-MARIE-TOOTH (CMT)

Definition of the disease

Charcot-Marie-Tooth (CMT) disease is the most prevalent peripheral inherited neuropathy (1/2500 to 10 000; 2.8/10 000 in Spain), and the mean age at onset is 16 years (range 2 to 50 years, but presentation in the early infancy and as late as the 80s has been reported). Patients present with motor and sensory polyneuropathic semiology (distal lower limb weakness and atrophy, gait abnormalities and frequent falls) and pes cavus. Apart from the motor nerve related deficits, most patients suffer slight sensory loss in hands and feet. The treatment of the disease is supportive. Life expectancy is not shortened - except in some forms of Déjerine-Sottas and severe forms of CMT-, but disabilities are the rule.

Diagnosis

The diagnosis is based on clinical and EMG/NCV findings, and in many instances by identification of diagnostic changes in one of the genes that determine the CMT2 subtypes.

Management and treatment

Treatment by a team including a neurologist, physiatrists, orthopedic surgeons, physical, and occupational therapist; special shoes and/or ankle/foot orthoses (AFO) to correct foot drop and aid walking; surgery as needed for severe pes cavus; forearm crutches, canes, wheelchairs as needed for mobility; exercise as tolerated; symptomatic treatment of pain, depression, sleep apnea, restless legs syndrome.

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