Limb-Girdle Muscular Dystrophy Awareness Day

Frequently Asked Questions

What is Limb-Girdle Muscular Dystrophy (LGMD)?

Limb-girdle muscular dystrophy (LGMD) is a general term used to describe a group of genetically inherited neuro-muscular diseases that cause weakness and wasting of the muscles around the shoulders and hips. Under the umbrella of LGMD there are more than 30 identified sub-types, with new forms being discovered each year.

Individuals with LGMD experience progressive weakness and wasting of the muscles closest to their body (proximal) such as the shoulders, upper arms, pelvic area, and thighs. The degree to which an individual's muscle ability is affected can vary from person to person and even between those who are part of the same sub-type or biological family. The progressive weakness of muscles can result in changes in posture and appearance. In certain LGMD sub-types, the heart muscle is weakened. Weakness in the muscles that support breathing can also result in breathing difficulties for some individuals. Symptoms of LGMD can be present early in life or develop later in life. Likewise, the progression of the condition can be rapid and debilitating to slow with limited degeneration. The common denominator for all individuals with LGMD is progressive muscle weakness and loss of related muscle function. LGMD occurs in all parts of the world & among all ethnic groups. Males and females can inherit LGMD

How many people have LGMD?

LGMDs are considered rare diseases. It is difficult to determine the prevalence of limb-girdle muscular dystrophy because its features vary and overlap with those of other muscle disorders. Estimates range from 1 in 14,500 to 1 in 123,00 individuals, depending on the sub-type. As more individuals obtain genetic testing to confirm their LGMD diagnosis, better estimates of prevalence may become available.

What causes LGMD?

Individuals with LGMD carry mutations in genes that provide faulty instructions for making proteins involved in muscle maintenance and repair. As there are many sub-types of LGMD there are also many different genetic mutations that underlie these conditions.

What are genes and how do they work?

Our bodies are made up of millions cells. Within the cells there are small thread-like structures called chromosomes, formed of DNA. Genes are located on the chromosomes and they provide a set of instructions for how our bodies grow and work. We have thousands of genes that provide information from eye color to muscle function. We inherit two copies of most genes, one from the father and one from the mother. In rare instances, a spontaneous mutation in a gene appears and is passed onto future generations.

How is LGMD classified?

LGMD is classified by its inheritance pattern and underlying genetic cause. Inheritance patterns are either autosomal dominant or autosomal recessive.

Autosomal Dominant: LGMD sub-types with a "1" in the nomenclature (such as LGMD1A, LGMD1B) have an autosomal dominant inheritance pattern. In autosomal dominant inheritance, only one parent needs to transmit the mutated gene for the disorder to be inherited. Even though the matching gene from the other parent is unaffected, the faulty gene dominates. Autosomal dominant disorders typically appear in every generation without skips.

If a person affected with an autosomal dominant disorder decides to have children, each offspring has a 50% chance of inheriting the faulty gene and developing the disorder. Children who do not inherit the affected gene will not develop or pass the disorder to their offspring.

Those individuals with an autosomal dominant pattern of LGMD are classified as "type 1." When the specific mutated gene is identified, a letter is then assigned to designate the specific LGMD sub-type. For example, an individual who has inherited a mutation in the CAV3 gene in a dominant pattern is identified as having LGMD Type 1C or LGMD1C.

Autosomal Recessive: LGMD sub-types with a "2" in the nomenclature (such as LGMD2A, LGMD2B) have an autosomal recessive inheritance pattern. In autosomal recessive inheritance, both parents must be carriers of the faulty gene for their child to inherit the disease. A carrier harbors the faulty gene, but usually shows no symptoms. When a mutation is recessive, it can exist in a person without showing any symptoms so long as their other copy of the same gene is healthy. The unaffected gene masks the mutation. However, if a child inherits two copies of the mutated gene, there is no healthy alternative to block the faulty gene out, and so the symptoms manifest themselves.

If both parents have one copy of the mutated gene, with each pregnancy, there is a 25% chance the child will receive a faulty gene from each parent and thus be affected by the disorder. If the child inherits a faulty gene from one parent only, he or she will be an unaffected carrier of the disorder. Autosomal recessive disorders typically appear when there is no known family history of the disorder

Those individuals with an autosomal recessive pattern of LGMD are classified as "type 2". When the specific genes are identified a letter is then assigned to designate the specific sub-type. For

example, an individual who has inherited a mutation in the CAP3 gene in a recessive pattern would be identified as having LGMD Type 2A or LGMD2A. .

Most forms of LGMD are inherited in a recessive (type 2) pattern.

Is there a cure?

Currently no cure or treatment exists for these progressive diseases.

Since the late 1990's there has been a large upswing in the number of research projects and papers on LGMD. Improved and more specific genetic testing has enabled individuals to obtain more detailed information about their sub-type. As more individuals with LGMD are associated with a specific sub-type researchers will have the opportunity to work with a defined group of individuals. Clinical trials are currently being conducted using gene therapy and medications with animal subjects and laboratory models. There are treatments options for individuals with certain manifestations of LGMD. For example, when the heart muscle is affected there are medications that can reduce the impact of the heart damage on the person's overall well-being. Getting a genetically confirmed sub-type is a critical step toward finding a cure.

What are some of the challenges experienced by individuals with LGMD?

Individuals with LGMD have difficulty obtaining an accurate diagnosis. Many individuals show early symptoms of unexplained muscle weakness leaving medical professionals with a wide array of possibilities when looking for a diagnosis. Until recent refinements in genetic testing, it was difficult, if not impossible to get an accurate diagnosis.

Limited mobility results in many challenges for individuals with LGMD from physical restrictions, to pain and social isolation. Living with a progressive, degenerative condition can impact an individual's emotional and psychological well-being. Those individuals who have sub-types affecting the heart or breathing may need full or part time ventilation support to breath and medication to ease the stress on the heart muscle.

A common misconception people have about LGMD is that it can be improved with more exercise. While exercise within the limits of an individual's condition is helpful to maintain range of motion and strengthen unaffected muscles, exercise will not cure LGMD and may (when exaggerated) actually harm existing muscles.

What is Limb-Girdle Muscular Dystrophy Awareness Day?

LGMD Awareness Day is a collaborative effort to globally raise awareness of individuals living with limb-girdle muscular dystrophy. This global event is celebrated annually on September 30th. LGMD Awareness Day is a project of the LGMD Awareness Foundation, Inc., a 501(c)(3) non-profit advocacy organization.

When is Limb Girdle Muscular Dystrophy Awareness Day?

"Limb-Girdle Muscular Dystrophy Awareness Day" is celebrated annually on September 30th worldwide. The first LGMD Awareness Day was celebrated in 2015.

Where is Limb-Girdle Muscular Dystrophy Awareness Day being held?

Our intention is that the Limb-Girdle Muscular Dystrophy Awareness Day will be celebrated and acknowledged globally. There won't be a single event, but instead we are encouraging people in many parts of the world to set up activities to commemorate the day, according to their customs and practices.

We are planning activities to raise awareness leading up to and around September 30th. Please visit our website at LGMD-Info.org to learn how to become an "Ambassador" for the awareness day in your regions. We will also be posting ideas on our Facebook page. And, of course, we welcome any ideas that you have.

Where can I get more information?

To obtain more information on LGMD, available LGMD organizations and resources as well as information on the upcoming Limb-Girdle Muscular Dystrophy Awareness Day, please visit our website at: **LGMD-Info.org** or email us at info@LGMD-Info.org

Also, consider following us on Facebook, Twitter and Instagram @LgmdAwareness

LGMD
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