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Anatomy and Physiology Article Critique

The article *Limb-girdle muscular dystrophy: What nurses need to know* by Kaitlyn Sahd, Lisa Ruth-Sahd, and Keith Brazzo, published in the Nursing2017 magazine, informs readers about the causes, symptoms, and treatments of a genetic muscle-destroying disease, abbreviated as LGMD. This inherited disorder is characterized by weakening of the most body-weight-bearing muscle structures, such as pelvic and shoulder girdles and calves. As a result, seemingly healthy children become unable to get up, walk, and even breathe.

As determined by genetic testing, LGMD is caused by a mutation of a gene encoding any protein necessary for muscle function. For example, if dystrophin, normally found in sarcoplasm, is absent, myofibrils get destroyed, and muscles atrophy. Most cases of LGMD are autosomal recessive, which means that LGMD carriers may be healthy themselves but pass the defected allele to their children. As of today, more than 20 forms of LGMD are discovered, based on genes encoding abnormal proteins. Boys and girls are equally at risk, and estimated 0.5 to 4 in 100,000 people get sick with LGMD worldwide.

The early signs and symptoms of LGMD are progressive muscle weakness, primarily in hips and shoulders, and difficulty getting up from the seated position. Autosomal recessive, or LGMD2 types, usually progress more rapidly, and affected children lose mobility by adolescence, and many die by adulthood of heart failure, because cardiac muscles also lose

tone. Luckily, though, smooth muscles are not affected, as stated in *Facts About Limb-Girdle Muscular Dystrophy* brochure by Muscular Dystrophy Association (MDA).

Treatment options for patients with LGMD include light to moderate physical exercise to improve strength and range of motion; swimming is especially recommended. Massage, hot baths, and stretching help loosen tight muscles. Universal recommendations to eat healthy, maintain an optimal body weight, and sleep enough also apply. Additionally, patients need to see specialists, including cardiologist, neurologist, and orthopedist regularly and be prepared for use of a wheelchair and a cane when weakness progresses.

The article effectively incorporates facts and expert opinions from twenty different sources, as specialized publications as online, for the last decade. For example, the author refers to the three credible research studies about the use of coenzyme Q-10 and L-glutamine to improve muscle function in patients with muscular dystrophy. The statistics on prevalence of LGMD among other muscle-destroying disorders and percentage of people affected are gathered from a peer-reviewed article by Basil Darras, professor of Neurology in Harvard Medical School. Recommendations are combined from the several sources, including those written by E. Pegoraro, PhD (Department of Neurosciences in University of Padova, Italy), E. Hoffman, PhD (Research Center for Genetic Medicine, Washington, DC), and other field experts. The author also mentions ongoing research studies involving gene therapy, myoblast transplantation, and other new treatments proposed by experienced neurologists P. Narayanaswami (Harvard Medical School) and others, published in Neurology magazine in 2014.

Although LGMD is rare, it is the fourth most common form of MD, after dystrophinopathies (Duchenne MD and Becker MD), myotonic dystrophy, and facioscapulohumeral MD. Because it

is so rare, parents do not expect that something so unlikely can happen to their child. We can only imagine how shocked parents must be upon discovering it in their family.

Questions:

1. Is there any prenatal DNA testing available, where both prospective parents can find out if any of them is a carrier of a disease-causing allele? Using biotechnology, can couples estimate chances of passing a genetic disease to their prospective children?

2. Has a similar genetic disorder been found in animals? If there is a similar disorder in mice, for instance, they could be used for new medication testing and gene therapy, which could help determine possible effects and risks for human patients.

3. More than 20 forms of LGMD are identified and encoded, such as several LGMD1 forms for autosomal dominant and several LGMD2 forms for autosomal recessive inheritance. Defected proteins are found in various parts of muscle cells: in sarcolemma, on either actin or myosin myofilaments, in nucleus, or in sarcoplasm, all having different functions in cells. Would it be better to discard LGMD abbreviation completely, and instead call all these cases by their names, based on the defected or missing proteins, on the first place? As I see, all forms of LGMD have only couple of traits in common: weakness in hips, waddling gait, and autosomal inheritance. Naming the various cases more descriptively may help target a cause more precisely and make treatment more effective.

Nowadays, medical experts conduct researches and develop new treatments almost rapidly, and children with LGMD can get adults, make careers, and have their own children, which is fascinating. No doubt, life expectancy of patients with muscular dystrophies is going to raise, and their quality of life will continue to be improving in the following years.

Works Cited

Sahd, Kaitlyn E. BSN, RN, CPN; Ruth-Sahd, Lisa DEd, RN, CEN, CCRN; Brazzo, Keith G. DPM. "Limb-girdle muscular dystrophy: What nurses need to know." Nursing2017 47.7 (2017): 54-58.