Marfan Syndrome

By Monica Lepore and Maria Lepore-Stevens

ETIOLOGY

Marfan syndrome (MFS) is an inherited, autosomal dominant condition caused by a mutation in a FBN1 gene, which affects the body's connective tissue (CDC, 2019). The mutation of the FBN1 gene interferes with the body's production of protein necessary for building connective tissue. Connective tissue provides structure and strength to many of our organ systems. Disordered connective tissue causes damage to a person's skin, lungs, eyes, heart, skeleton, and blood vessels, and other structures. Another name for this syndrome is contractural arachnodactyly (Genetic and Rare Diseases Information Center [GARD], 2017).

GENERAL CHARACTERISTICS

Characteristics of Marfan syndrome (MFS) are dependent upon what parts of the body are affected and how severely the person is affected. Some individuals show little to no characteristics while others have many of the more than 80 possible symptoms. Structurally, many individuals with MFS have an elongated face; lean and tall body structure; long fingers, toes, and arm span; sunken or protruding chest; hyperflexible joints; flat feet; arthritis; and scoliosis or kyphosis (National Heart, Lung and Blood Institute [NHLBI], 2019). Some individuals with MFS also have stretch marks on their skin, lung disorders (spontaneous lung collapse), and abdominal or inguinal hernias. Abnormalities related to the heart and aorta include: leakage in the heart valves, irregular heartbeat (feelings like skipping a beat or palpitations), increased size of aorta with aortic valve leakage and possible aortic dissection, and aortic aneurysm. Aortic dissection and aortic aneurysm are life threatening (NHLBI, 2019).

VISUAL IMPAIRMENT AND MARFAN SYNDROME

Most individuals with Marfan syndrome live with high myopia (extreme nearsightedness) and astigmatism (atypical curvature of the lens of the eye). Approximately 60% of individuals with MFS have ectopia lentis, a dislocation of the lens which could result in progressive vision loss over time (American Association for Pediatric Ophthalmology and Strabismus [AAPOS], 2020). Marfan syndrome is also associated with a higher incidence of glaucoma in mid-adulthood, cataracts, esotropia (inward turning eye), or exotropia (outward turning eye), and retinal detachment (GARD, 2017). Approximately 5-29% of individuals with MFS have an increased risk of retinal detachment, which can cause total blindness (GARD, 2017; National Organization for Rare Disorders [NORD], 2021).

SPECIFIC MODIFICATIONS IN A UNIVERSALLY DESIGNED PHYSICAL EDUCATION LESSON

- Create safe squads for all participants who need low impact activities and areas that lack flying projectiles.

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- Announce three levels of intensity for all aerobic activities. Give guidelines for the variety of levels, including rest intervals.
- Provide heart rate watches to a variety of students, including the individual with MFS.
- Provide incentives for all to concentrate on skill-form rather than time, distance, and intensity.
- Create a choice for individual skill practice versus all-out full-sided sports and games.
- Utilize light-weight foam balls for throwing and catching.
- Provide time during warm-ups when all students can communicate how they are feeling that day, such as having students give a thumbs up, sideways, or down during student check-in.
- Allow students to opt out of high-intensity or contact activities and have an alternate activity that focuses on the skills of the activity, always planned and set up in a safe area.

CONTRAINDICATIONS

Individuals with MFS have some physical activity considerations, especially those with cardiac conditions and risk of retinal detachment. Students who have an identified risk of retinal detachment may need to avoid activities in which they could get hit in the head, such as head-first diving in the pool, wrestling, and full-game contact sports (NORD, 2021). Although more research is needed on ideal intensity, frequency, duration, and type of exercise for people with MFS, the current recommendations are for mild to moderate aerobic exercise that is low intensity and low impact (Jensen et al., 2020). Functionally, this means the individual can participate in continuous aerobic movement at a pace in which they can carry on a conversation. Current research also advocates tight cardiac monitoring, or monitoring of blood pressure, heart rate, and perceived exertion, in communication with cardiologist (Jensen et al., 2020). The Marfan Foundation (2017) recommends aerobic levels of about 50% of capacity. These exercise guidelines must be further adapted for cardiac or other medications that affect heart rate or blood pressure. Recommendations for physical activity are not the same from person to person, as each individual comes to the table with a variety of characteristics. Working with the individual, their family, and their physician, is imperative in the MFS population (Marfan Foundation, 2017). The chart on page 7 of the Marfan Foundation Physical Activity Guidelines detail specific activities that are high, moderate, and low risk (see reference list). The Marfan Foundation (2017) also recommends that students with MFS avoid the following activities:

- Exercise that causes the individual to grunt, hold their breath, bear down, or strain to levels of exhaustion.
- Collision and contact sports
- Exercise testing at max levels or exercise to exhaustion
- Yoga or gymnastics poses that put excessive stress on the body such as headstands and shoulder stands
- Intense isometric exercises
- Activity intensity of greater than 6 METS (energy equivalent of expending 6 times the energy you would at rest). Some examples of activities that are above 6 METS are: singles tennis, team basketball, team soccer, hiking uphill, bicycling or swimming at a fast pace

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SPECIFIC RESOURCES

Pennington, C. G., & Webb, L. M. (2020) Enhancing physical education for students with vision impairment and preventing retinal detachment. *Journal of Physical Education, Recreation & Dance*, 91(3), 53-54. DOI: 10.1080/07303084.2019.1705134

Jensen, T. L., Tran, P., & Kjaer, M. (2020). Marfan syndrome and exercise: A literature review. *Translational Sports Medicine*, 3(6), 526-535. https://doi.org/10.1002/tsm2.185

Webb, L. M. & Pennington, C. G. (2019) Facilitating Physical Activity with Individuals with Marfan Syndrome, *Journal of Physical Education, Recreation & Dance*, 90(8), 64-65. DOI: 10.1080/07303084.2019.1649573

REFERENCES

American Association for Pediatric Ophthalmology and Strabismus. (2020). *Marfan syndrome*. https://aapos.org/glossary/marfan-syndrome

Centers for Disease Control and Prevention. (2019). *Marfan Syndrome*. https://www.cdc.gov/heartdisease/marfan_syndrome.htm

Genetic and Rare Diseases Information Center (GARD). (2017). *Marfan syndrome*. U.S. Department of Health & Human Services, National Institutes of Health. https://rarediseases.info.nih.gov/diseases/6975/marfan-syndrome

Marfan Foundation. (2017). *Physical activity guidelines*. https://info.marfan.org/hubfs/FINAL%20Physical%20Activity%20Guidelines%2011_17.pdf

National Heart, Lung and Blood Institute. (2019). *Marfan syndrome*. U.S. Department of Health & Human Services, National Institutes of Health. https://www.nhlbi.nih.gov/health-topics/marfan-syndrome

National Organization for Rare Disorders. (2021). *Marfan syndrome*. https://rarediseases.org/rare-diseases/marfan-syndrome

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