Appendix 1

Prompt #1

Write 10 questions regarding Duchenne Muscular Dystrophy for an interview with a medical professional expert. Answer those questions in formal, scientific language, each answer should have about 400-500 words. Cite sources.

Output #1

Q1. What is Duchenne Muscular Dystrophy (DMD)?

A1. Duchenne Muscular Dystrophy (DMD) is a genetic disorder caused by mutations in the dystrophin gene, which is responsible for producing the protein dystrophin. Dystrophin is a protein found in muscle cells that helps keep them strong and functioning properly. Without dystrophin, the muscles become weak and are unable to function properly. DMD is a progressive disorder, meaning that the symptoms worsen over time, and it is the most common form of muscular dystrophy in children. It is estimated to affect 1 in every 3,500 to 5,000 male births worldwide (1).

Q2. What are the symptoms of DMD?

A2. The symptoms of DMD vary depending on the age of the individual, but generally include muscle weakness, 1contractures, difficulty breathing, and cardiac abnormalities. As the disease progresses, the individual may experience difficulty standing, difficulty swallowing, and eventually require the use of a wheelchair. In some cases, individuals may also experience cognitive impairment, seizures, and scoliosis (2).

Q3. What are the causes of DMD?
A2. DMD is caused by mutations in the dystrophin gene, which is located on the X chromosome. The dystrophin gene is responsible for producing the protein dystrophin, which is necessary for muscle cells to function properly. Mutations in this gene cause the body to produce an abnormal or non-functioning dystrophin protein, which leads to the muscle weakness and other symptoms associated with DMD (3).

Q4. How is DMD diagnosed?

A4. DMD is typically diagnosed through a combination of clinical examination, genetic testing, and muscle biopsy. During the clinical examination, a doctor will look for signs of muscle weakness, joint contractures, and other physical abnormalities. Genetic testing can be used to identify mutations in the dystrophin gene, and a muscle biopsy can be used to confirm the diagnosis. In some cases, other tests such as an electromyogram (EMG) or a nerve conduction study may be used to help diagnose DMD (4).

Q5. What treatments are available for DMD?

A5. Currently, there is no cure for DMD, but there are a number of treatments available to help manage the symptoms and slow the progression of the disease. These treatments include physical therapy, occupational therapy, speech therapy, and medications to help manage muscle weakness, joint contractures, and other symptoms. In some cases, surgery may be used to help improve mobility and function. Additionally, individuals with DMD may benefit from the use of assistive devices such as wheelchairs and walkers (5).

Q6. What is the prognosis for individuals with DMD?

A6. The prognosis for individuals with DMD varies depending on the severity of the disease and the age at which it is diagnosed. Generally, individuals with DMD have a shorter life expectancy than those without the disorder. Most individuals with DMD will require the use of a wheelchair by the age of 12, and the average life expectancy is around 25 years (6).

Q7. Are there any clinical trials for DMD?

A7. Yes, there are a number of clinical trials currently underway for DMD. These trials are investigating potential treatments such as gene therapy, stem cell therapy, and medications to help manage the symptoms of DMD. Additionally, there are a number of clinical trials that are looking at ways to improve the quality of life for individuals with DMD (7).

Q8. Are there any support groups or resources available for individuals with DMD?

A8. Yes, there are a number of support groups and resources available for individuals with DMD. These include local support groups, online support groups, and national organizations such as the Muscular Dystrophy Association (MDA). Additionally, there are a number of websites and blogs dedicated to providing information and support for individuals with DMD (8).

Q9. What research is being done to find a cure for DMD?

A9. Currently, there is a great deal of research being done to find a cure for DMD. This research includes gene therapy, stem cell therapy, and medications to help manage the symptoms of DMD. Additionally, researchers are looking at ways to improve the quality of life for individuals with DMD, such as developing assistive devices and improving physical therapy and occupational therapy techniques (9).

Q10. What advice would you give to parents of a child with DMD?

A10. The most important advice I would give to parents of a child with DMD is to seek out support and resources. It is important to connect with other families who have a child with DMD, as well as organizations such as the Muscular Dystrophy Association (MDA). Additionally, it is important to stay informed about the latest research and treatments available for DMD, and to make sure that your child is receiving the best possible care. Finally, it is important to remember that each child is unique and to focus on the positive aspects of their life, such as their strengths and abilities (10).

References:

1. "Duchenne Muscular Dystrophy (DMD)." Muscular Dystrophy Association, [www.mda.org/disease/duchenne-muscular-dystrophy](http://www.mda.org/disease/duchenne-muscular-dystrophy).

2. "Duchenne Muscular Dystrophy." National Institute of Neurological Disorders and Stroke, U.S. Department of Health and Human Services, [www.ninds.nih.gov/disorders/duchenne/detail\_duchenne.htm](http://www.ninds.nih.gov/disorders/duchenne/detail_duchenne.htm).

3. "Duchenne Muscular Dystrophy." Genetics Home Reference, U.S. National Library of Medicine, ghr.nlm.nih.gov/condition/duchenne-muscular-dystrophy.

4. "Duchenne Muscular Dystrophy." Mayo Clinic, Mayo Foundation for Medical Education and Research, [www.mayoclinic.org/diseases-conditions/duchenne-muscular-dystrophy/diagnosis-treatment/drc-20374519](http://www.mayoclinic.org/diseases-conditions/duchenne-muscular-dystrophy/diagnosis-treatment/drc-20374519).

5. "Duchenne Muscular Dystrophy." Muscular Dystrophy Association, [www.mda.org/disease/duchenne-muscular-dystrophy/treatment](http://www.mda.org/disease/duchenne-muscular-dystrophy/treatment).

6. "Duchenne Muscular Dystrophy." National Institute of Neurological Disorders and Stroke, U.S. Department of Health and Human Services, [www.ninds.nih.gov/disorders/duchenne/detail\_duchenne.htm#3019\_5](http://www.ninds.nih.gov/disorders/duchenne/detail_duchenne.htm#3019_5).

7. "Clinical Trials for Duchenne Muscular Dystrophy." ClinicalTrials.gov, U.S. National Library of Medicine, [www.clinicaltrials.gov/ct2/results?cond=Duchenne+Muscular+Dystrophy](http://www.clinicaltrials.gov/ct2/results?cond=Duchenne+Muscular+Dystrophy).

8. "Resources for Duchenne Muscular Dystrophy." Muscular Dystrophy Association, [www.mda.org/disease/duchenne-muscular-dystrophy/resources](http://www.mda.org/disease/duchenne-muscular-dystrophy/resources).

9. "Research for Duchenne Muscular Dystrophy." Muscular Dystrophy Association, [www.mda.org/disease/duchenne-muscular-dystrophy/research](http://www.mda.org/disease/duchenne-muscular-dystrophy/research).

10. "Tips for Parents of a Child with Duchenne Muscular Dystrophy." Muscular Dystrophy Association, [www.mda.org/disease/duchenne-muscular-dystrophy/tips-for-parents](http://www.mda.org/disease/duchenne-muscular-dystrophy/tips-for-parents).

Prompt #2

Write a scientific paper on Duchenne Muscular Dystrophy focusing on treatment advances in formal, scientific language. Cite sources in [the] Vancouver citation style.

Output #2

Introduction

Duchenne muscular dystrophy (DMD) is an X-linked recessive genetic disorder characterized by progressive muscle degeneration and weakness. It is the most common form of muscular dystrophy, affecting approximately 1 in 5,000 live male births (1). The disorder is caused by a mutation in the dystrophin gene, which results in the absence of dystrophin, a protein that is essential for muscle function (2). The lack of dystrophin leads to progressive muscle degeneration, resulting in a variety of symptoms including muscle weakness, contractures, and cardiomyopathy (3). The disorder is typically diagnosed in early childhood and is associated with a shortened lifespan (4). Despite the severity of the disorder, advances in treatment have been made in recent years, offering hope for those affected by DMD.

Treatment Overview

The primary treatment for DMD is corticosteroid therapy, which is used to reduce muscle inflammation and slow the progression of the disorder (5). Other treatments include physical therapy, orthopedic surgery, and cardiac care (6). Additionally, gene therapy has been explored as a potential treatment for DMD, though it is still in the early stages of development (7).

Corticosteroid Therapy

Corticosteroid therapy is the primary treatment for DMD and is used to reduce muscle inflammation and slow the progression of the disorder (5). The most commonly used corticosteroid is prednisone, which has been shown to improve muscle strength and function in those with DMD (8). Additionally, long-term use of corticosteroids has been associated with a reduced risk of scoliosis, cardiomyopathy, and respiratory complications (9).

Physical Therapy

Physical therapy is an important component of DMD treatment and is used to improve muscle strength and function (10). Physical therapy can also help to reduce the risk of contractures and scoliosis (11). Additionally, physical therapy can help to improve balance and coordination, as well as reduce fatigue (12).

Orthopedic Surgery

Orthopedic surgery is sometimes used to treat DMD, particularly for those with severe contractures or scoliosis (13). The most common surgeries used to treat DMD are tendon lengthening and joint fusion (14). These surgeries can help to improve range of motion and reduce pain (15).

Cardiac Care

Cardiac care is an important component of DMD treatment, as the disorder is associated with an increased risk of cardiomyopathy (16). Cardiac care typically includes regular monitoring and medication to reduce the risk of complications (17).

Gene Therapy

Gene therapy is a promising treatment for DMD, though it is still in the early stages of development (7). The goal of gene therapy is to replace the mutated dystrophin gene with a healthy version, which could potentially restore muscle function (18). While gene therapy is still in the early stages of development, it has shown promise in animal models and clinical trials are currently underway (19).

Conclusion

Duchenne muscular dystrophy is a severe genetic disorder characterized by progressive muscle degeneration and weakness. While the disorder is associated with a shortened lifespan, advances in treatment have been made in recent years, offering hope for those affected by DMD. Treatment typically includes corticosteroid therapy, physical therapy, orthopedic surgery, cardiac care, and gene therapy. While gene therapy is still in the early stages of development, it has shown promise in animal models and clinical trials are currently underway.

References

1-7 and 16-19. Kinali, M., & Bushby, K. (2015). Duchenne muscular dystrophy. The Lancet, 386(9995), 829-841.

8-15. Mendell, J. R., & Rodino-Klapac, L. R. (2015). Corticosteroid therapy for Duchenne muscular dystrophy. Muscle & Nerve, 51(2), 175-182.

*\* Repeated references were edited and put together.*

Prompt #3.1

How is respiratory failure treated in patients with Duchenne Muscular Dystrophy?

Output #3.1

Respiratory failure in patients with Duchenne Muscular Dystrophy is treated with a combination of medications, oxygen therapy, and mechanical ventilation. Medications such as bronchodilators and steroids may be used to reduce airway inflammation and improve airway function. Oxygen therapy can help increase oxygen levels in the body, while mechanical ventilation can help stabilize breathing and reduce the risk of respiratory infections. In some cases, surgery may be necessary to improve airway function or to insert a breathing tube.

Prompt #3.2

Explain Respiratory management strategies in Duchenne Muscular Dystrophy

Output #3.2

Respiratory management strategies in Duchenne Muscular Dystrophy (DMD) involve a combination of medical, physical, and psychosocial interventions. Medically, bronchodilators and steroids may be used to reduce airway inflammation and improve airway function. Oxygen therapy can also be used to increase oxygen levels in the body. Physically, regular exercise and postural drainage techniques can help maintain pulmonary function. Psychosocially, psychological support can help manage the emotional and cognitive effects of the condition. Additionally, respiratory rehabilitation programs may be employed to provide education and training on how to better manage shortness of breath and other symptoms.

Prompt #3.3

What is the role of mechanical ventilation in respiratory management strategies in Duchenne Muscular Dystrophy?

Output #3.3

The role of mechanical ventilation in respiratory management strategies for Duchenne Muscular Dystrophy is to help stabilize breathing and reduce the risk of respiratory infections. Mechanical ventilation can also provide respiratory support during periods of acute respiratory distress, and can be used to help clear secretions from the lungs. In some cases, mechanical ventilation may be necessary to maintain adequate oxygen levels in the body, and to prevent complications such as respiratory failure. In addition, mechanical ventilation may also be used to provide respiratory support for patients who cannot breathe effectively on their own.

Prompt #3.4

What are Respiratory management strategies including the role of nocturnal assisted breathing and mechanical ventilation in Duchenne Muscular Dystrophy?

Output #3.4

Respiratory management strategies for Duchenne Muscular Dystrophy (DMD) involve a combination of medical, physical, and psychosocial interventions. Medically, bronchodilators and steroids may be used to reduce airway inflammation and improve airway function. Oxygen therapy can also be used to increase oxygen levels in the body. Physically, regular exercise and postural drainage techniques can help maintain pulmonary function. Psychosocially, psychological support can help manage the emotional and cognitive effects of the condition. Additionally, respiratory rehabilitation programs may be employed to provide education and training on how to better manage shortness of breath and other symptoms. Nocturnal assisted breathing devices such as continuous positive airway pressure (CPAP) machines and bi-level positive airway pressure (BiPAP) machines can help stabilize breathing during sleep and reduce the risk of respiratory infections. Mechanical ventilation may also be necessary to maintain adequate oxygen levels in the body and to prevent complications such as respiratory failure.

Prompt #4

Please summarize the following article: […]

Whole main text of the article was passed to the prompt, Abstract, Figures, Tables, Limitations and Conclusions were not included.

Shehta M, Rayan MM, Fahmy NA, Onsy A, Bastawy I. Global longitudinal strain detects subtle left ventricular systolic dysfunction in Duchenne muscular dystrophy patients and carriers. Egypt Heart J. 2021 Oct 19;73(1):91. doi: 10.1186/s43044-021-00214-0. PMID: 34665363; PMCID: PMC8526669.

Output #4

This study aimed to detect subtle cardiac changes in Duchenne muscular dystrophy (DMD) patients and carriers using electrocardiography (ECG) and echocardiography. A cross-sectional case-control study was conducted of 28 male DMD patients and 25 female carriers compared to gender-matched healthy control groups. Results showed smaller left ventricular (LV) wall diameters, LV end-diastolic diameter (LVEDD), left atrium (LA) diameter and worse global longitudinal strain (GLS) in the DMD and carrier groups compared to the control groups. Intra- and inter-observer variability in strain analysis was acceptable. The study found that GLS is a useful tool for early detection of subtle LV systolic dysfunction, allowing the early start of anti-failure treatments that may improve prognosis.

Output #4 Factcheck

Aim – correct

Study dosing – correct

Sample definition – correct

Results:

 the DMD **and** carrier groups compared to the control groups

smaller left ventricular (LV) wall diameters – WRONG for DMD, TRUE for carriers

LV end-diastolic diameter (LVEDD) – TRUE for both

left atrium (LA) diameter – TRUE for both

worse global longitudinal strain (GLS) – TRUE for both

*no mention of higher heart rates, smaller LV end-systolic diameter and lower ejection fraction in DMD*

 Intra- and inter-observer variability in strain analysis was acceptable - correct

Conclusions – correct