

# Duchenne Muscular Dystrophy

## CARRIERS

Parent  
Project  
Muscular  
Dystrophy

### What Does Being a Carrier Mean?

A carrier of Duchenne muscular dystrophy (Duchenne) is typically a female (XX) with one altered copy of the dystrophin (DMD) gene, located on the X chromosome. This genetic change may be inherited from a parent or occur spontaneously (de novo variant). While many carriers do not experience symptoms, they do have an increased risk for heart disease, and can have additional physical or psychological impacts.

### What Are the Health Risks for Carriers?

- **Heart:** Female carriers are at an increased risk of developing cardiomyopathy (a weakening of the heart muscle) or arrhythmias (irregular heartbeats).
- **Muscle:** Some carriers experience difficulty with physical activities such as climbing stairs or running. They may also have muscle cramps, pain, and fatigue easily. In rare cases, muscle symptoms progress over time and are significant enough to be called muscular dystrophy.
- **Emotional/Psychological:** Carriers may experience psychological impacts related to the risk of passing on the condition or developing symptoms, and they have higher rates of learning and behavioral differences related to the genetic variant.

### Health Monitoring and Care for Carriers

To manage potential health risks, carriers should take a proactive approach:

#### 1 Cardiac Monitoring

- Begin heart evaluations by early adulthood and repeat testing as recommended by a cardiologist familiar with Duchenne.
- Testing includes heart imaging (echocardiogram/MRI) and screening for arrhythmia (EKG)

#### 2 Neuromuscular Care

- If muscle symptoms (weakness, cramping) develop, consult a neuromuscular specialist.
- Physical therapy or other interventions may be recommended for managing symptoms.

#### 3 Family Planning Support

- Make an appointment with a genetic counselor to learn about the risk of Duchenne for other family members.

#### 4 Mental and Emotional Well-being

- Connect with counseling or peer support groups to address concerns and reduce stress related to being a carrier.
- If a learning or behavioral disability is suspected, meet with a psychiatrist for an evaluation.

## Genetic Testing for Carrier Status

Genetic testing identifies variants in the dystrophin gene to determine if someone is a carrier.

**Who Should Be Tested?** Testing can be considered for all pregnant people or those planning a pregnancy, and especially for:

- Individuals with a family history of Duchenne.
- Mothers of children with Duchenne.
- Women with unexplained muscle weakness or cardiac symptoms.

Testing is a simple blood or cheek swab test and can provide crucial information for health management and family planning.

## Reproductive Considerations

Carriers have a 50% chance of passing the altered gene to their children:

- **Sons:** Inheriting the variant leads to Duchenne muscular dystrophy.
- **Daughters:** Inheriting the variant leads to health risks associated with carrier status

Options to reduce transmission risk include IVF and other means of growing a family, such as adoption.

## PPMD Support and Resources

PPMD is dedicated to providing carriers with tools, knowledge, and connections to manage their health and improve their quality of life:

- **Carrier Research:** Ongoing studies explore the unique health needs of carriers, focusing on cardiac, muscular, and emotional well-being.
- **Support Programs:** PPMD offers support groups, educational webinars, and resources tailored to carriers and their families.
- **Advocacy:** PPMD works to ensure carriers have access to appropriate healthcare, insurance coverage, and genetic counseling.

## Empowering Carriers to Take Charge of Their Health

Carriers play a crucial role in advancing the understanding of Duchenne and ensuring better outcomes for future generations. By staying informed, undergoing regular health evaluations, and seeking support, carriers can lead healthy, fulfilling lives.

# CONTACT PPMD FOR MORE INFORMATION

Together, we can ensure every carrier has the resources and care they need.



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