Jett FOUNDATION

Empowering families.
Fighting Duchenne.

# Female with Dystrophinopathy or DMD Manifesting Carrier

## JOURNEY TO DIAGNOSIS & NEXT STEPS

### THE JOURNEY TO DIAGNOSIS

It's important to know that not just adults or moms to children who have been diagnosed with Duchenne can be carriers or symptomatic females. Any first-degree female relative such as a grandmother, sister, aunt, or even cousin can be diagnosed.

Alternatively, no family history is even required. A female could have what is called a spontaneous mutation, meaning the cause for the mutation is unknown. This is being seen in more pediatric girls who are symptomatic. Approximately 30% (1 out of 3) of children born with Duchenne have a genetic change that started new in them and was not inherited from their mother (PPMD).

It is not yet known how common heart changes are, but some studies have estimated that 10-50% of carriers have heart changes (PPMD). Symptoms to be aware of and to look out for in all ages, especially if there is a known family history. (However, please note, that just like with any rare disease, it is important to acknowledge that everyone is different. There can be ranges of having no symptoms at all, to being severely impacted, and anything in between):

- Elevated creatine kinase (CK) levels, which are found through a blood test. This is one of the first signals to do genetic testing for Duchenne.
- Fatigue or inability to be physically active for long periods, may have difficulty walking or running for long distances, or difficulty climbing stairs or jumping. May need extended time to recover from illnesses.
    - Pay close attention to signs of cardiac involvement such as shortness of breath, feeling of flutters or extra beats, or feeling like heart skipping beats, dizziness, really fast heart rate, just generally feeling of not feeling well.
- Muscle pain, enlarged calf muscles

If you suspect your child may be a carrier or if you have a family history of Duchenne, **genetic testing to confirm status** is very important and should be step one after seeing symptoms or learning of family diagnosis. If you feel like it is right for your family, test at an early age if you suspect your child is a suspected carrier or if you have a history of Duchenne in your family. Once confirmed, it's time to compile resources and to build a care team.

## NEXT STEPS AFTER DIAGNOSIS

*Please note, at this time, there are no official next steps after being diagnosed as a Carrier or Female with Dystrophinopathy. These are recommendations based on the experience of those in the community, and should be used to help guide others in their journey.*

**You are your own best advocate.** Being able to advocate for yourself is extremely important. There is still a lot of misinformation in the community about x-linked disorders, specifically carrier information, and how they affect females in the Duchenne space. Access to knowledgeable clinics and physicians is also not currently widespread.

### *Begin building your Care Team*

- Speak with your primary care physician about your diagnosis and learn about their willingness and ability to help guide you through your journey and work with other physicians who are knowledgeable. If the one you currently have won't, reach out to others and the community to help.
- If you have a son with Duchenne, work with their current care team to determine if they will be willing to treat you or your daughter.
- This care might not be available locally, you may have to travel once or twice yearly to clinics who are knowledgeable in carrier care.
  - Please visit **Jett Foundation's website** to learn more about providers and clinics that are knowledgeable and willing to treat females with Duchenne. This list is not extensive and is actively being built. If you have a name to add, please reach out to info@jettfoundation.org.

### *Request a Referral to a Cardiologist*

- Bring your genetic test results with you and any other medical information that may be important to share
- Important Cardiac tests and imaging needs - Again, please note that these are based on recommendations that carriers in the community have received in their care, and there is not currently a set of standards of care for females:
  - EKG and echo at baseline and yearly.
  - If pediatric and symptomatic, individuals should get a Cardiac MRI by age 13 for baseline and every 2-3 years after that.
  - For adults, you should receive a baseline MRI at the time of diagnosis to check for fibrosis (the thickening and scarring of connective tissue, usually as a result of injury), and every 3-5 years after that, unless recommended for a shorter timeline by your care team.

### *Depending on your symptoms and needs, request referrals, as needed, for pulmonology, physical medicine, physical therapy, occupational therapy, etc.*

- Not all symptomatic carriers will have muscle issues, some may have only cardiac issues, and some may have a combination of both.
- Dystrophinopathies cause a wide spectrum of manifestations in affected individuals, from males with DMD and BMD to manifesting female carriers. It is a multisystemic disease, which necessitates multidisciplinary specialized care.

### *If ready and willing, connect with support groups in the Duchenne community:*

**Jett Foundation Family Support Groups:**

- **Carrier Conversations:** Carrier Conversations is Jett Foundation's monthly virtual support group for women in the Duchenne community who are carriers of Duchenne muscular dystrophy (whether they are manifesting or not). This group is a safe space for carriers to connect and chat with each other. It is an informal, intimate setting that ensures women feel comfortable and empowered in organic conversations that arise, which may include but are not limited to: care, being a caregiver, self-care, medical issues, guilt, and more.
- **Caregiver Connextions:** Caregiver Connexions is Jett Foundation's monthly virtual support group for parents, family members or caregivers of females who have been diagnosed with Duchenne or are symptomatic carriers. This group is a safe space for caregivers (moms, dads, aunts, uncles, grandparents, guardians, etc.) to connect and chat with each other about caring for their daughters, granddaughters, and female loved ones impacted by Duchenne. It is an informal, intimate setting that ensures caregivers feel comfortable and empowered in organic conversations that arise, which may include but are not limited to: care, being a caregiver, self-care, supporting siblings, guilt, and more.

### *Do your research.*

- Go to the Jett Foundation's list of **Resources for Female Carriers** to learn more about the current information, research articles and resources out there for carriers. These are always being updated and as we receive more information, we will continue to add them here.