

# Caring for Duchenne Carriers

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**Project** END DUCHENNE.  
**Muscular  
Dystrophy**

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# What is a DMD carrier?

- Female with presence of DMD mutation, located at Xp21.2
- Frequency:
  - DMD frequency of 1:5000 live male births
  - Mom is somatic carrier in 2/3 of cases
  - Remaining 1/3 *de novo* or due to germline mosaicism (~10%)

# What is a manifesting carrier?

- Traditionally thought to be DMD carrier who exhibits spectrum of symptoms seen in DMD
- Symptoms ranging from mild to severe
- Often thought to be due to skewed X-chromosome inactivation

# What we know, and what we don't

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- One study suggested that 2.5-7.8% of carriers will manifest musculoskeletal symptoms
- Only 1/3 of cohort had skewed XCI
- 80% had weakness, 60% had myalgias
- Age at onset of symptoms from 2-47 years

# What we know, and what we don't

- Dilated Cardiomyopathy prevalence
  - 8-18% per Kondo et. al
  - 3-84% per Childers et. al
  - 36-84% per Viggiano et. al

# What we know, and what we don't

- Study of 182 moms via DuchenneConnect
  - 75% knew their carrier status, 25% did not
- Half of those who did not know their status attributed this to financial barriers
- Majority of moms who didn't know carrier status didn't understand risk of having another son with DMD

# What we know, and what we don't

- Clear disparity between data
- Need for increased education and access to services
- Minimal literature regarding psychological burden of being a carrier while caring for an affected son
- Need more studies to better characterize

# Current Care of Carriers

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- All mothers of children with DMD/BMD should be offered carrier testing
  - Own care considerations
  - Future pregnancies
  - Other children
- Continued access to genetic counseling regardless of carrier status

# Current Care of Carriers

- AAP consensus meeting in 2009:
  - Education on risk of developing DCM and symptoms of heart failure
  - Referral to cardiologist in late adolescence or early adulthood
  - Rescreening every five years starting at 25-30 years of age
  - Treated with similar approach as DMD DCM

# Current Care of Carriers

- Assessment of myalgia, weakness, fatigue
- Limitation of physical abilities, integration of PT/OT
- Need for intermittent, regular monitoring
- Pain management modalities

# Current Care of Carriers

- Psychosocial assessment
- Role of social work
- Need for care/skilled nursing services at home to help in care of DMD sons

# Improving Carrier Care

- Knowledge of carrier status and implications
- Home care needs
- Group support in community
- Linking to resources

# We Need More Information!

- There's still much to learn about the manifestations and needs of our carrier moms
- PPMD-sponsored study evaluating carrier and non-carrier moms
- Eric Camino, PhD, CCRC with interim data

# Questions?

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