Caring for Duchenne Carriers

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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
What we know, and what we don’t

• 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
Current Care of Carriers

• 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?
References


