X-linked Myotubular Myopathy: Overview of Genetics

Essential Information:
- *MTM1* gene found on long arm of the X chromosome
- Inheritance: X-linked recessive manner

Finding a gene mutation is important for:
- Accurate diagnosis
- Prognosis (what to expect)
- Carrier detection & other family members
- Family planning

Why do some female carriers of *MTM1* mutations have symptoms?
- 80-90% chance a woman is a carrier and her son inherited the genetic change from her
- 10-20% chance the genetic change was *de novo* (new) in the son and not inherited from mom

*MTM1* Genetic Testing Laboratories

<table>
<thead>
<tr>
<th>Laboratory</th>
<th>Phone</th>
<th>Website</th>
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<tbody>
<tr>
<td>The University of Chicago Genetic Services Lab</td>
<td>888.824.3637</td>
<td>dnatesting.uchicago.edu</td>
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<tr>
<td>Prevention Genetics</td>
<td>715.387.3637</td>
<td>preventiongenetics.com</td>
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<tr>
<td>Emory Genetic Laboratory</td>
<td>404.778.8499</td>
<td>genetics.emory.edu/egl</td>
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If you have additional questions or are interested in more information about our research studies, please contact me:

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