

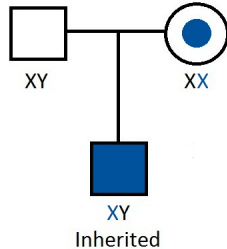


# X-linked Myotubular Myopathy: Overview of Genetics

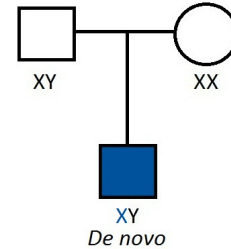


## Essential Information:

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



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

## Finding a gene mutation is important for:

- Accurate diagnosis
- Prognosis (what to expect)
- Carrier detection & other family members
- Family planning
- Prenatal diagnosis or PGD (preimplantation genetic diagnosis)
- Development of targeted treatments and cures

## Why do some female carriers of *MTM1* mutations have symptoms?

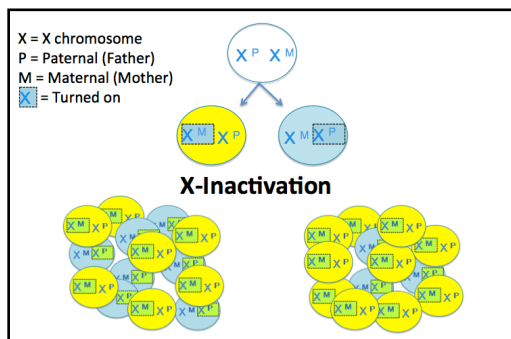
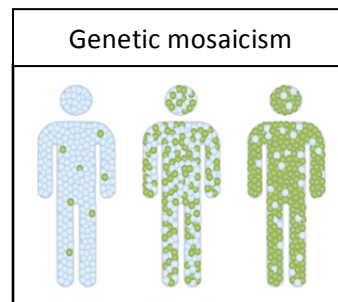


Image courtesy of Elizabeth DeChene, MS, CGC



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## *MTM1* Genetic Testing Laboratories

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| The University of Chicago Genetic Services Lab | 888.824.3637 | <a href="http://dnatesting.uchicago.edu">dnatesting.uchicago.edu</a> |
| Prevention Genetics                            | 715.387.3637 | <a href="http://preventiongenetics.com">preventiongenetics.com</a>   |
| Emory Genetic Laboratory                       | 404.778.8499 | <a href="http://genetics.emory.edu/egl">genetics.emory.edu/egl</a>   |

If you have additional questions or are interested in more information about our research studies, please contact me:

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