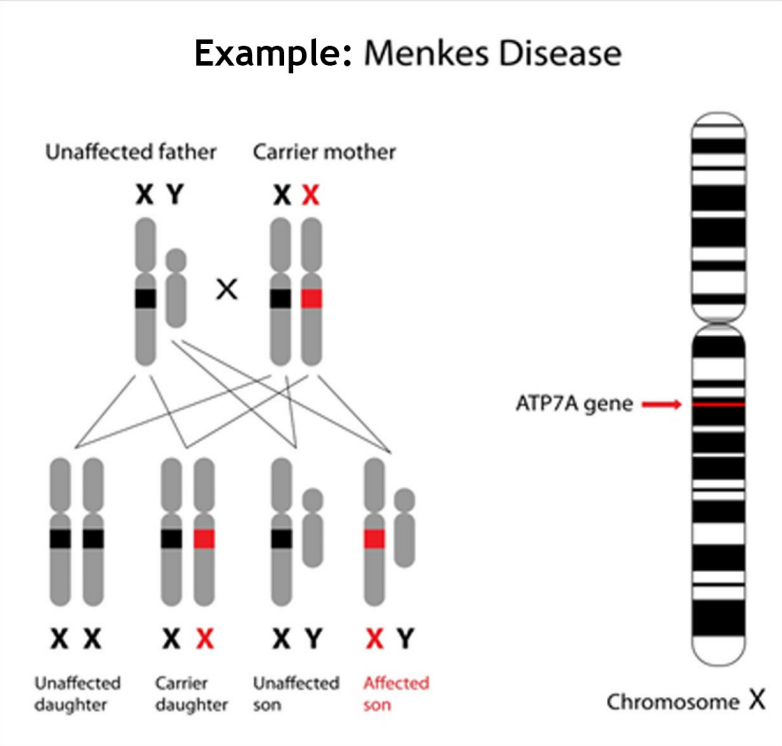
2.1.4 What Causes Genetic Conditions?



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Menkes disease is a recessive genetic condition caused by a mutation of the gene on the X chromosome that is responsible for the metabolism of copper in the body. Copper levels become abnormally low in the liver and brain and abnormally high in the kidney and intestinal lining. Menkes disease leads to premature birth, floppy muscle tone, developmental delay, seizures, and failure to gain weight and to grow at the usual rate.[1,2,3]

Gene mutations can be inherited from parents; however, mutations in genes and chromosomes can happen unexpectedly and usually for unexplained reasons. Mutations that exist in the parents' genes are inherited mutations. New mutations, which occur during the transmission of genetic material to the next generation, are called de novo mutations. It is important to obtain a complete family history to better understand genetic risk factors for children and their families.

For more information about inheritance see:

- SickKids, 'Genetics' section (If a user profile box pops up, you can press 'cancel' or create a user profile to access this site)
- Centers for Disease Control and Prevention, 'Pediatric Genetics' section

1 http://ghr.nlm.nih.gov/condition/menkes-syndrome;

2 http://www.merriam-webster.com/medical/menkes'%20disease;

3 http://www.ninds.nih.gov/disorders/menkes/menkes.htm