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MITOCHONDRIAL DISORDERS

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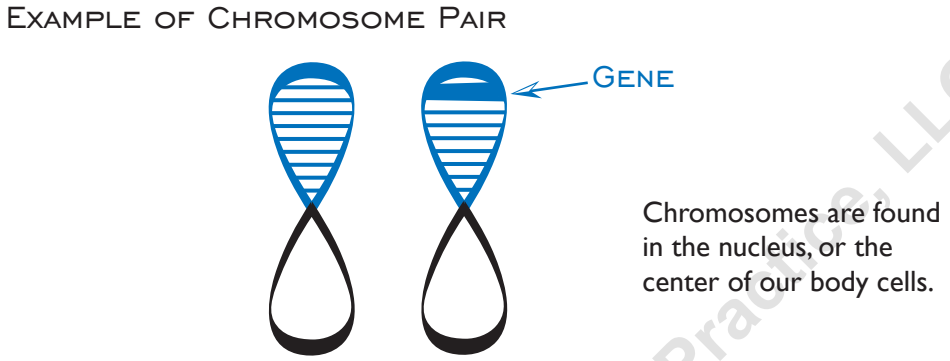
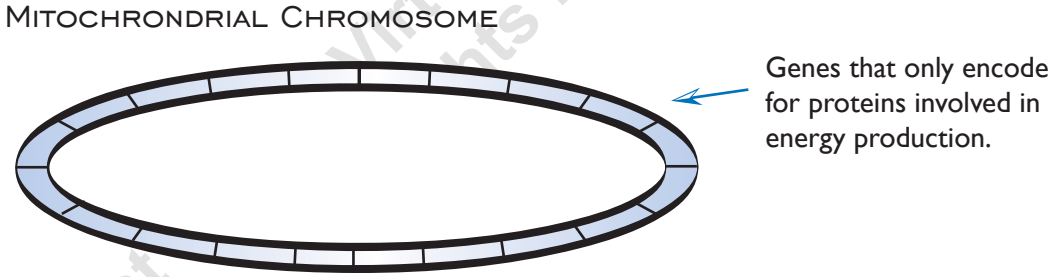
Mitochondrial Disorders

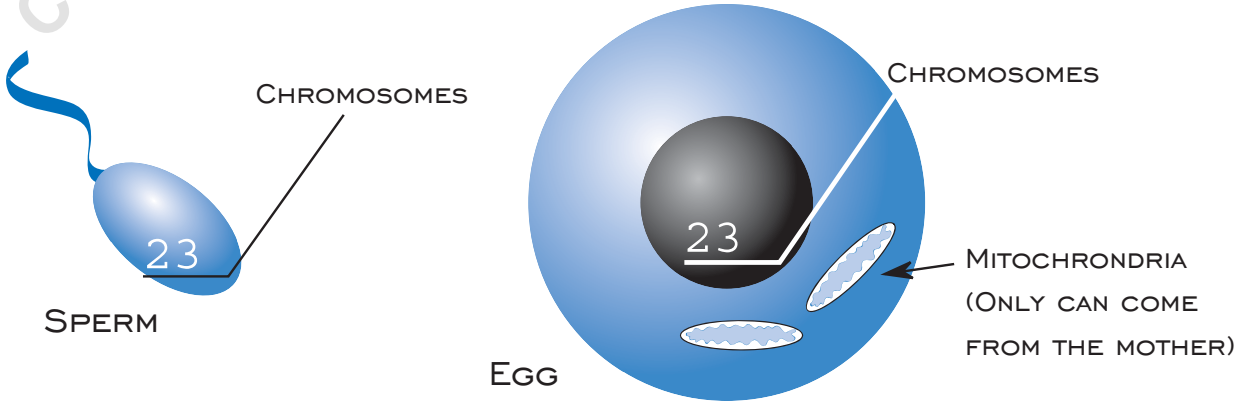
Human beings have 46 chromosomes, 23 or ½ of which are inherited from each parent at the time of conception. Chromosomes are found in the nucleus, or center, of our body cells. The body cell is the smallest functioning unit of the body. A cell is like a small town and the nucleus like city hall. Genes, or individual units of heredity are located on chromosomes. Each gene encodes for some protein.

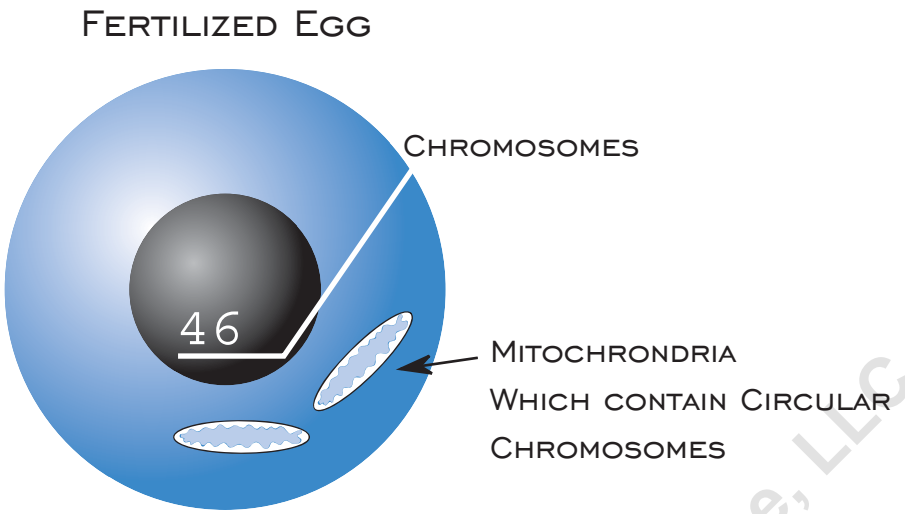


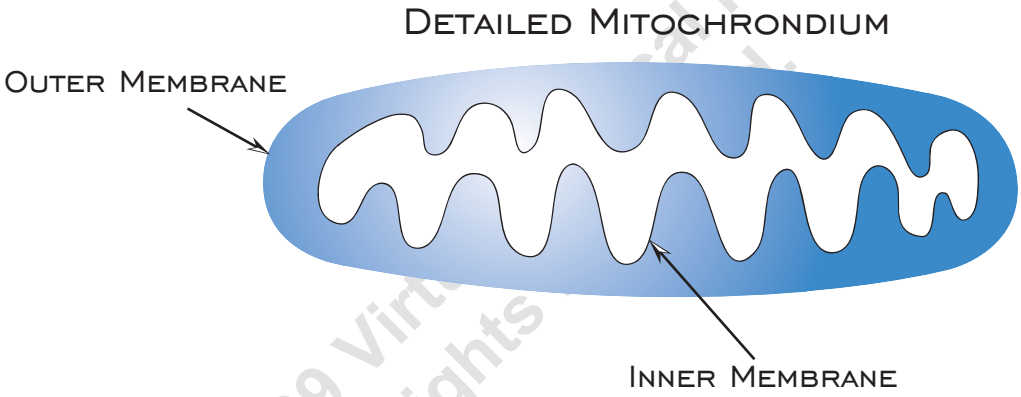
We also inherit large circular chromosomes only from our mother. These genes only encode for proteins involved in energy production. These chromosomes are located inside our mitochondria, the "batteries" or electric companies of our cells, providing energy for body functions.



A new human being gets all of this genetic material at conception.

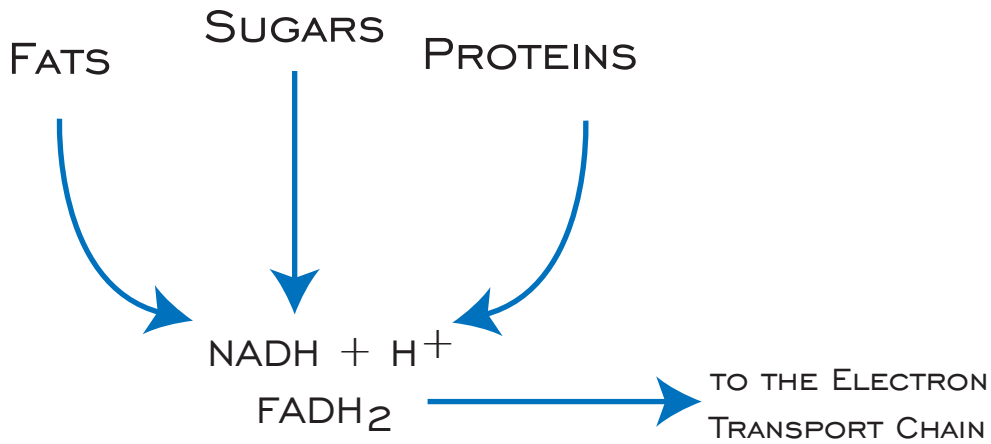




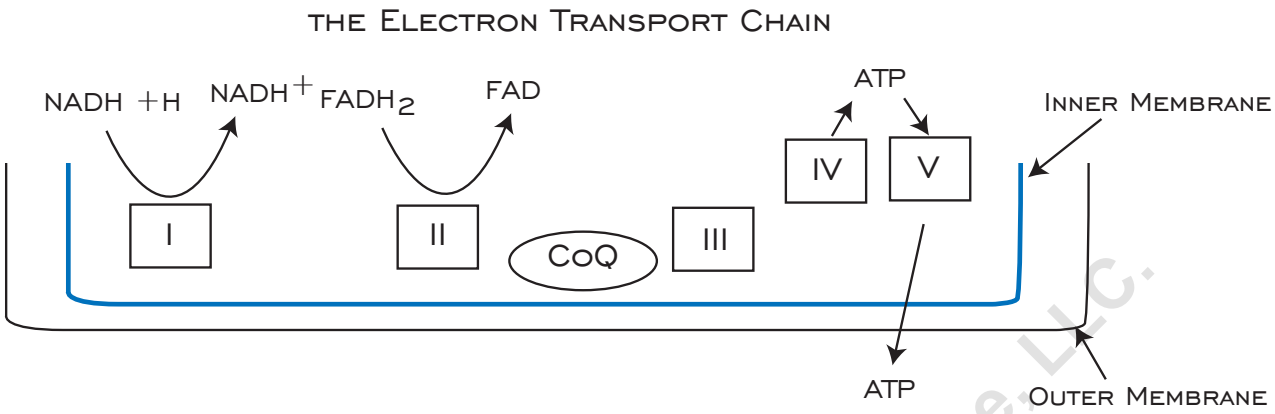


Embedded within the mitochondrial inner membranes are the five complexes of the electron transport chain. Each complex is made up of many proteins. All of these proteins work together to produce energy packets known as ATP. This process is known as oxidative phosphorylation. ATP is a universal form of energy used throughout the body for any number of tasks.

When food is broken down inside our body cells, this process creates high energy electrons that are shuttled to the mitochondria for final processing.



The electrons are dumped into the electron transport chain and ATP is produced.



Mitochondrial disorders occur when there is a mutation or change in either one of the genes in the nuclear or mitochondrial chromosomes. These genes include those that make up the proteins that:

1. *are found in one of the complexes*
2. *transport certain proteins into the mitochondria*
3. *assemble the proteins together to allow them to work normally.*

Because we cannot look at all the genes that cause mitochondrial disorders we must examine the enzyme system (electron transport chain) that produces ATP by taking a piece of tissue rich in mitochondria (muscle).

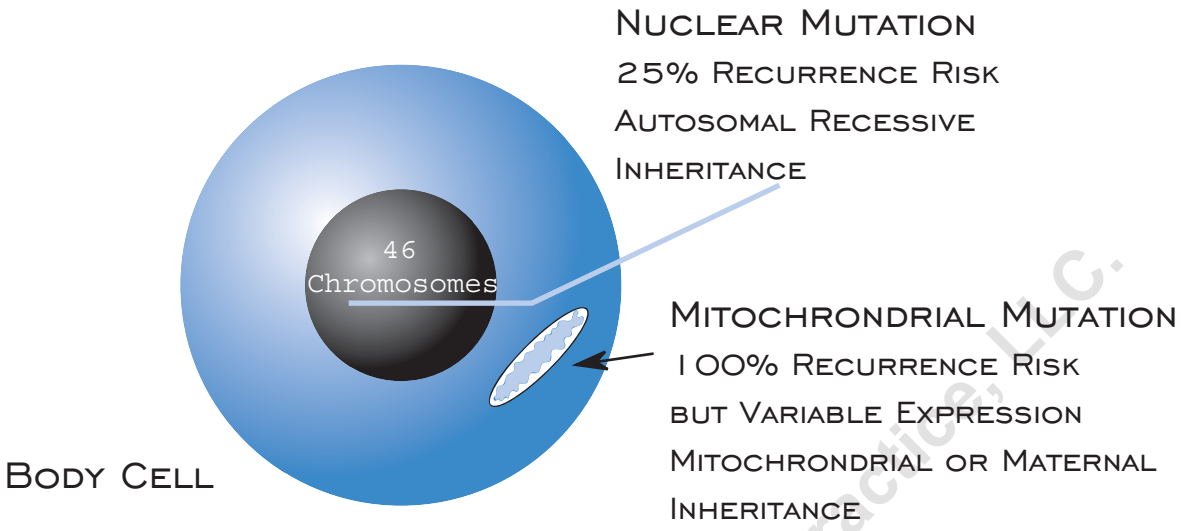
Patients affected with these disorders display or are at risk for developing multi-system problems. The body systems most affected by decreased energy production are:

1. *central nervous system (brain)*
2. *heart*
3. *muscle*
4. *kidney*
5. *liver*

No one person will necessarily develop difficulties with all these body systems but all are at risk for such problems as they advance through their lives. Other problems include diabetes, thyroid disease and gastrointestinal difficulties.

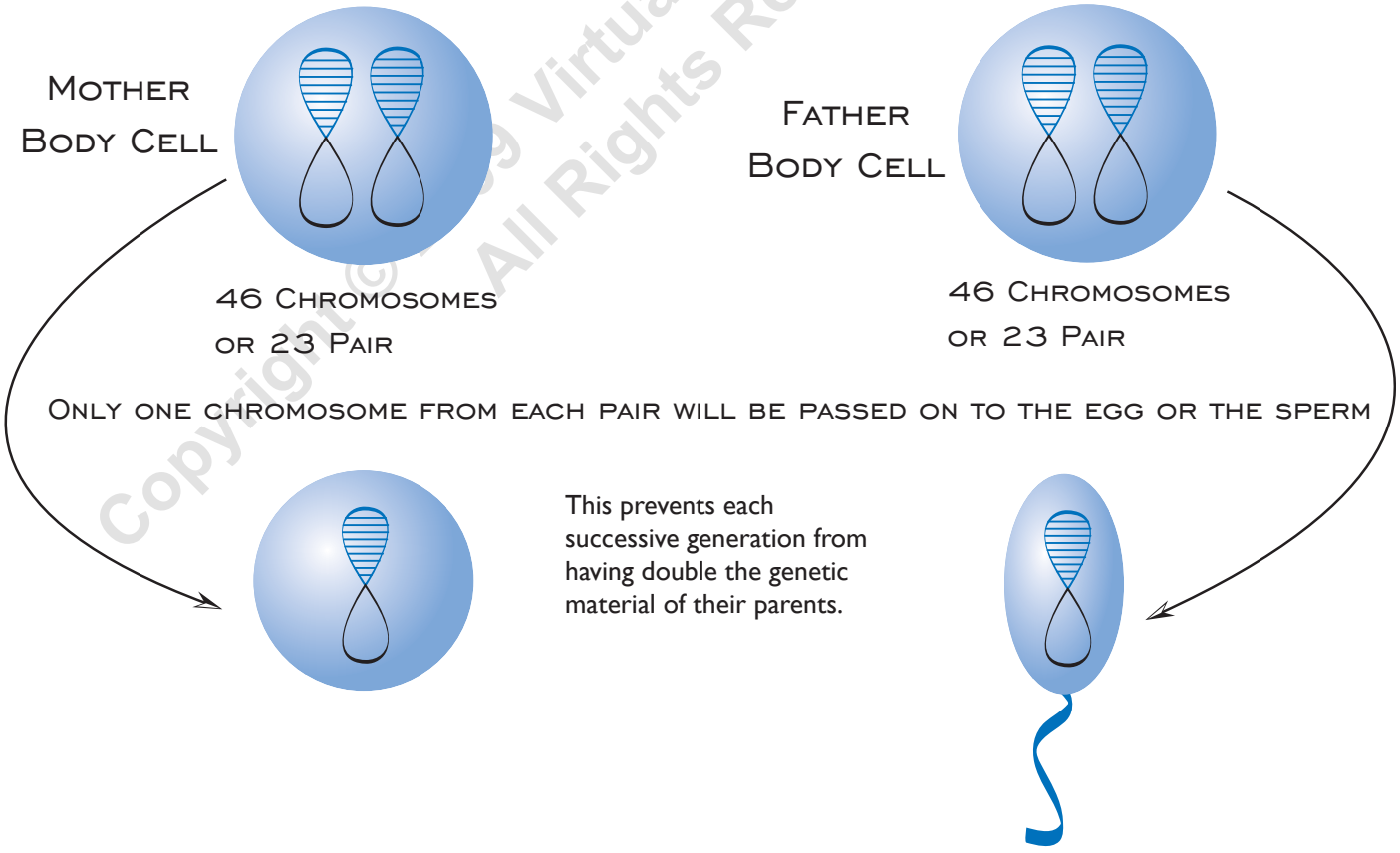
There is no cure for these disorders. Treatment is symptomatic (for example, if you have seizures you receive anticonvulsants) and preventative. A number of problems can occur that can be treated early, such as diabetes, so a yearly screen for these difficulties is recommended. The use of some supplements, including Coenzyme Q_{10} has been shown to be beneficial for some patients.

Mitochrondrial disorders are genetic disorders. Genetic disorders are inherited, so couples with one affected child are at risk for having other children with the same disease. The recurrence risk depends on whether or not the changed gene was nuclear or mitochondrial.

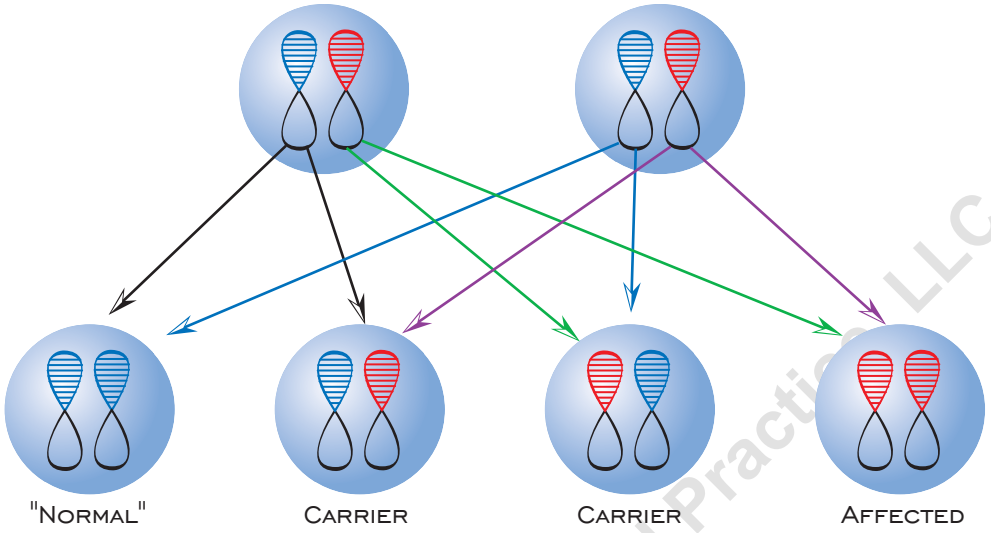


Autosomal Recessive Inheritance

Eggs and sperm contain ½ of a parent's genetic material.

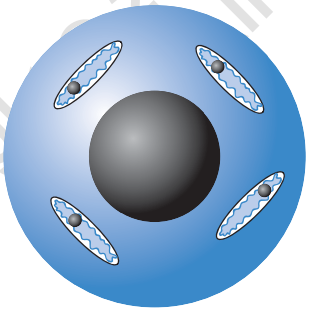


If a parent carries a change or mutation in one chromosome in a given pair, he or she will pass it on to the offspring 50% of the time. However, since most genes must have a change in both members of a pair to cause a problem, a child is affected only if he or she inherits a changed gene from each parent.



Maternal Inheritance

If a mother carries a changed or mutated mitochondrial gene, she will pass it on to her children via the egg. The amount of changed mitochondrial DNA found in a fertilized egg will determine how affected a new baby will be. If many mutated copies are found, most of the body tissues formed from that one cell (fertilized egg) will be affected. If few copies are found, few symptoms or problems may develop.



Fertilized Egg

with Many Mutated Copies

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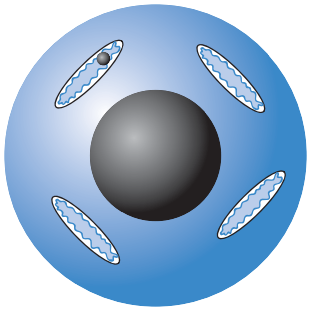
Severe Problems



Fertilized Egg

with Few Mutated Copies

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Few or No Problems