



Genetics and muscular dystrophy

Families impacted by neuromuscular diseases eventually ask the same question: How did this happen to my child or spouse? Genetic diseases are seemingly random but scientifically are easily explained. Many people have asked me questions about genetic diseases and the following information is helpful to understand genetic diseases.

Chromosomes come in pairs in the cell's nucleus. Humans have 46 chromosomes in each cell nucleus, which are actually 23 pairs of chromosomes. One chromosome comes from a person's mother, the other from his father. For 22 of these pairs, numbered chromosome 1 through chromosome 22, the chromosomes are the same; that is, they carry genes for the same traits.

The 23rd pair is an exception and determines gender. The 23rd chromosomal pair differs according to whether you're a male or a female. Males have an X and a Y chromosome, while females have two Xs for this 23rd pair of chromosomes. Every female gets one X chromosome from her mother and one X from her father. Every male gets an X chromosome from his mother and a Y from his father.

Diseases covered by MDA fall into two types: autosomal dominant and autosomal recessive.

Autosomal dominant conditions require only one mutation to show themselves. In autosomal dominant disorders, the chance of having an affected child is 50 percent with each conception.

Autosomal recessive conditions require two mutations to show themselves. When the term "autosomal recessive" is used, it means that the disorder is again located on chromosomes that aren't X's or Y's. However, when a disorder is recessive, it takes two mutated genes to cause a visible disorder in a person.

The word "recessive" comes from the idea that, when only one gene mutation exists, it may remain undetected ("recede" into the background) for several generations in a family—until someone has a child with another person who also has a mutation in that same autosomal gene. Then, the two recessive genes can come together in a child and produce the signs and symptoms of a genetic disorder.

You can think of recessive genes as "weaker" than "dominant" genes, in that it takes two of them to cause a problem.

People with one gene mutation for disorders that require two to produce the disorder are said to be carriers of the disorder. Carriers are usually protected from showing symptoms of a genetic disease by the presence of a normal corresponding gene on the other chromosome of each chromosome pair.

In autosomal recessive disorders, the chance of having an affected child is 25 percent with each conception.

The diagrams (at right) are mathematical calculations of the odds that one gene or the other in a pair of genes will be passed on to a child during any particular conception.

These are the same kinds of calculations one would make if asked to predict the chances of a coin landing as heads or tails. With each coin toss (assuming the coin isn't weighted and the conditions are otherwise impartial), the chances that the coin will land in one position or the other are 50 percent.

In reality, if you were to toss a coin six times, you might come up with any number of combinations: All your tosses might be heads, or five could be heads with one tails, or four might be tails with two heads.

Parents of children sometimes feel guilty about their role in passing on the gene that causes muscular dystrophy. The MDA counselors who work with them make sure they know it is not their fault. Knowledge about how genetics work helps them cope with a situation that is beyond anyone's ability to control. ☒

