People of Eastern European (Ashkenazi) Jewish heritage can trace their ancestry to a “bottleneck” of just 350 individuals, dating back to between 600 and 800 years ago. Because the Ashkenazi community started out so small, and remained genetically isolated, it developed a higher load of disease-causing mutations. Even today, Ashkenazi Jews are known to be at higher risk for many diseases (all autosomal recessive):

* [**Bloom syndrome**](http://www.webmd.com/hw-popup/bloom-syndrome)**:** Babies with this disease are born small and remain shorter than normal as they grow. Their [skin](http://www.webmd.com/skin-problems-and-treatments/picture-of-the-skin) may look red, and they have more lung and [ear infections](http://www.webmd.com/cold-and-flu/ear-infection/default.htm) than children normally have.
* [**Canavan disease**](http://www.webmd.com/hw-popup/canavan-disease)**:** This disease gradually destroys [brain](http://www.webmd.com/brain/picture-of-the-brain) tissue.
* [**Cystic fibrosis**](http://www.webmd.com/hw-popup/cystic-fibrosis)**:** This disease causes very thick mucus in the [lungs](http://www.webmd.com/lung/picture-of-the-lungs) and problems with digesting food.
* [**Familial dysautonomia**](http://www.webmd.com/hw-popup/familial-dysautonomia) **(FD):** People with this problem cannot feel pain, they sweat a lot, and they have trouble with speech and coordination.
* [**Fanconi anemia**](http://www.webmd.com/hw-popup/fanconi-anemia)**:** People with this problem do not have enough [blood cells](http://www.webmd.com/heart/anatomy-picture-of-blood) and have problems with the [heart](http://www.webmd.com/heart/picture-of-the-heart), [kidneys](http://www.webmd.com/urinary-incontinence-oab/picture-of-the-kidneys), arms, or legs. They also are more likely to get [cancer](http://www.webmd.com/cancer/default.htm).
* [**Gaucher disease**](http://www.webmd.com/hw-popup/gaucher-disease)**:** This disease causes a type of fat called glucocerebroside to build up in certain cells of the [liver](http://www.webmd.com/digestive-disorders/picture-of-the-liver), [spleen](http://www.webmd.com/digestive-disorders/picture-of-the-spleen), and bone marrow.
* [**Mucolipidosis IV**](http://www.webmd.com/hw-popup/mucolipidosis-iv)**:** This problem causes the nervous system to deteriorate, or break down, over time.
* [**Niemann-Pick disease**](http://www.webmd.com/hw-popup/niemann-pick-disease) **(type A):** This disease causes a type of fat called sphingomyelin to build up in cells of the [liver](http://www.webmd.com/hepatitis/ss/slideshow-surprising-liver-damage), spleen, lymph nodes, and bone marrow.
* [**Tay-Sachs disease**](http://www.webmd.com/hw-popup/tay-sachs-disease)**:** This disease causes a type of fat called ganglioside to build up in the cells of the [brain and nervous system](http://www.webmd.com/brain/default.htm).
* [**Torsion dystonia**](http://www.webmd.com/hw-popup/torsion-dystonia)**:** People with this problem have ongoing spasms that twist the muscles in their arms, legs, and sometimes their body. Testing for this condition may not always be done.

About 1 out of 4 people of Ashkenazi Jewish heritage is a carrier of one of these genetic conditions. The most commonly carried alleles are for [Gaucher disease](http://www.webmd.com/a-to-z-guides/gauchers-disease-symptoms-causes-treatments), [cystic fibrosis](http://www.webmd.com/hw-popup/cystic-fibrosis), Tay-Sachs, familial dysautonomia, or [Canavan disease](http://www.webmd.com/children/canavan-disease-11143).

In situations today where both partners are of Ashkenazi descent, genetic testing is often sought prior to starting a family. Some of these diseases result in the child’s early death (for example Tay-Sachs can lead to death by age 4), thus by understanding the risks involved parents can make informed decisions. This is a classic example of when a dihybrid cross, or even a trihybrid cross may be appropriate, as parents may be carriers of two or more of these diseases.

**RULE OF MULTIPLICATION:** Independent events occurring in sequence (“and”)

**RULE OF ADDITION:** Mutually exclusive events (“or”)

1. A woman and her husband are both of Ashkenazi Jewish descent.  Both undergo genetic testing and discover that they are both carriers for cystic fibrosis AND Tay-Sachs disease. Determine the following probabilities for this couple:
2. Chance of having a healthy child (neither disease)?
3. Chance of a child with cystic fibrosis?
4. Chance of a child with cystic fibrosis or Tay-Sachs disease?
5. Chance of a child with both cystic fibrosis and Tay-Sachs disease?
6. Chance of a child who is a carrier for cystic fibrosis or Tay-Sachs disease?
7. Another couple, both of Ashkenazi Jewish descent undergoes genetic testing. The couple’s genotypes are as follows:

|  |  |  |
| --- | --- | --- |
| **Disease** | **Mother** | **Father** |
| Fanconi anemia | Carrier | Homozygous dominant |
| Niemann-Pick disease | Carrier | Carrier |
| Gaucher disease | Homozygous recessive | Carrier |

1. What is the chance that this couple may pass on Fanconi anemia to their child?
2. What is the chance that this couple will have a healthy child?
3. What is the chance that this couple will have two healthy children in a row?
4. What is the chance that this couple will have a child with Niemann-Pick or Gaucher.
5. How would the above information impact your family planning if you were one of the partners from #1 or #2? Go online and read more about these diseases before you write your response on a separate sheet of paper.