

Syndrome is between 1/9 and 1/12

Solving the Problem for Chances Are?

 Hb^{A} = normal beta globin

 Hb^{S} = sickle cell syndrome

Shaded symbols represent individuals who have been confirmed to have the disease sickle cell syndrome. Unshaded symbols represent people known not to have the disease. Squares are males. Circles are females. Individuals with sickle cell syndrome are healthy at birth but develop symptoms usually within the first year of life. Symptoms include <u>anemia</u>, which may result in decreased stamina; extreme <u>susceptibility to infections</u> (including septicemia [blood infection], pneumonia, bone infections and meningitis); <u>pain</u> (bone and many other body parts); <u>stroke</u> (rare in children); digestive duct <u>blockage</u> and right quadrant <u>pain</u> (such as the common bile duct).

The syndrome is inherited as an **autosomal recessive**. This means that both boys and girls will be affected and that any individuals who is Hb^{S}/Hb^{S} (using the gene symbols above) will have the disease. With adequate medical care, almost all affected individuals would be diagnosed with the disease in childhood. Therefore, any individual over about 1 year old who does not have symptoms of sickle cell syndrome (an unaffected individual with an unshaded symbol) is going to be either Hb^{A}/Hb^{A} or Hb^{A}/Hb^{S}

The rules of inheritance can be used to help determine if an unaffected individual is Hb^A/Hb^A or Hb^A/Hb^S . Sometimes it is possible to decide with certainty. Other times a probability (or chance) can be assigned to each of these two possibilities for an unaffected individual.

This is done in the above pedigree. For example, Scott's parents must both be Hb^A/Hb^S , called <u>carriers</u> of sickle cell, because they have an affected daughter. This also allows us to calculate Scott's chances of being a carrier as two out of three (2/3). Since Scott does not have the disease himself, the other possibility is that he is Hb^A/Hb^A with the remaining one out of three chance (1/3).

There is more uncertainty about Kim's chance of being a carrier, since her mother Mimi could be either Hb^A/Hb^A or Hb^A/Hb^S . Without more information about Mimi's family or her health status, it is not possible to assign her to one of the above types. Therefore she is either Hb^A/Hb^A , or Hb^A/Hb^S .

However, we can use each of Mimi's possibilities, <u>separately</u>, to calculate the "<u>range</u>" of probability that Kim and Scott will have a child affected with the disease. These are shown below using one of the Laws of Probability.

The overall probability of <u>two or more</u> independent events <u>all happening</u> is the product of each of the individual probabilities.

If Mimi is Hb^A/Hb^A , then Kim has a 1/2 chance of being a carrier.

| Therefore: | 1/2 | Х | | 2/3 | Х | | 1/4 | = 1/12 |
|------------|----------|------------------|-------|-----------------|-----------------|---------|---------------------------|--------|
| (Kim | 's chanc | e of being a car | rier) | (Scott's chance | e of being a ca | arrier) | (chance that two carriers | S |
| | | | | | | | will have an affected c | hild) |

If Mimi is Hb^A/Hb^S , then Kim has a 2/3 chance of being a carrier.

Therefore:2/3X1/4= 1/9(Kim's chance of being a carrier)(Scott's chance of being a carrier)(chance that two carriers
will have an affected child)