

## **How is EB Inherited?**

- Dominant Inheritance
- Recessive Inheritance
- Spontaneous Mutation

Most people with EB have inherited it through mutations in genes from their parents. Everyone has two copies of most genes: one copy from their mother and one from their father. Genes are in the body's cells and determine inherited traits such as eye color and dimples. They also govern some bodily functions, such as the formation of proteins in the skin. EB is caused by the abnormal formation or absence of these proteins. More than 10 genes are known to affect the formation of proteins in the skin. The type of EB someone has depends on which of these genes has a mutation and which protein has not been formed correctly.

### **Dominant Inheritance**

In dominantly inherited EB, the mutated gene is from only one parent who has the disease. When a disorder is dominantly inherited, only one faulty gene is required for the disorder to occur. A parent with a dominant form of EB has a 50-50 chance with each pregnancy that the baby will have EB. Birth order and sex of the child do not affect inheritance. If the abnormal gene is not passed to the child, the child will not have EB and cannot pass on the disorder.

### **Recessive Inheritance**

In recessively inherited EB, both parents carry an abnormality or mutation in the same gene. But because recessive traits and disorders require two copies of the gene for the trait or disorder to be present, neither of the parents has EB. Instead, the parents are carriers of this abnormal gene. In these parents, the second copy of the gene is normal. In order for EB to occur in their children, both parents must pass the mutated copy of the gene to the child. When two copies of the abnormal gene are paired, recessively inherited EB occurs. If only one parent passes the abnormal gene to the child, then the child will be a carrier but will not have EB. With recessive inheritance, there is a 25 percent chance with each pregnancy that the child will have the disorder. An individual with a recessive form of EB will be at risk of having an affected child only if he or she has a child with a carrier or another person with recessive EB.

### **Spontaneous Mutation**

EB also can be acquired through a new or spontaneous mutation. That is, a child is born with a dominant genetic mutation that causes EB, despite neither parent having EB. When this occurs, it is because the gene spontaneously mutated in either the sperm or the egg before conception. Once the child has a dominant gene for EB, he or she has a 50 percent chance of passing the disorder to his or her children, as explained above. There is no scientific evidence that the parents could have prevented such spontaneous mutation, and there are no known environmental, dietary or behavioral triggers for this type of mutation.