Epidermolysis Bullosa – Genetic Implications

Epidermolysis Bullosa (EB) occurs in approximately 1 in every 20,000 live births (or about 200 new children affected each year). Abnormalities in the proteins that holds the skin together is the cause of EB.

- The inherited forms of EB are genetic disorders, meaning the problem starts in the genes in the cells in the body. A gene is like a blue-print or a program that shows how the proteins that make up our cells and tissues are made.
- If the information is bad, then the protein won't work correctly. In the case of EB, the faulty genes lead to proteins that don't hold the layers of the skin together well enough and causes blisters/sores on the skin.

Type of Disorders

In general, every protein in the body has a set of two genes. One gene comes from the mother and one comes from the father.

Dominant Disorders

- Only one gene from one parent needs to be abnormal to produce the disease and be passed on to the child from the parent who has EB.
- More common than the recessive forms.
- Less severe and have fewer complications.

Recessive disorders

- Caused by abnormalities in both genes of the set. The child gets one abnormal gene from the mother and one abnormal gene from the father.
- Both parents are carriers for EB, but the parents do not have blisters, sore or other complications associated with EB.
- In recessive disorders, both parents must be carriers, and both genes the child gets must be faulty for the child to have symptoms of EB.
- The recessive forms are more severe and can have many complications.

There are four main forms of inherited EB:

- EB Simplex
- Junctional EB
- Dystrophic EB
- Kindler Syndrome

Chances of having another child with EB

It is important for a parent of a child with EB to know his or her chance of having another child with EB for future pregnancies. The chance varies with the different forms of EB.

Dominant inheritance

- If a parent has a dominantly inherited form of EB, there is a 50% chance with each pregnancy that the baby will be born with the same type of EB that the parent has.
- There is also a 50% chance that the child won't be born with that type of EB. If they are not born or show symptoms of EB, they will not be a carrier of it either.
- If a child has a dominantly inherited form of EB and neither parent has EB, the chances of having another affected child are very low. In these cases, the child has EB because of a “spontaneous” mutation of the abnormal gene.

Recessive inheritance is harder to understand.

- Parents who have a child with a recessively inherited form of EB don't have an EB diagnosis but carry the gene for it.
    - These parents often don't know that they are carriers of the gene until the baby is born. However, with each future pregnancy there is a 25% chance that their next baby will also be born with EB.
    - There is a 50% chance that their baby may be a carrier of it (like their parents) but not show the symptoms of EB.

Doctors will need to gather genetic information from both the child who has the EB diagnosis along with their parents to provide an accurate explanation of the risk for having another child with the same diagnosis.

Genetic counseling is strongly advised for:

- Families who have a child with an EB diagnosis and are thinking about having more children.
- Individuals with EB who are considering having children.

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