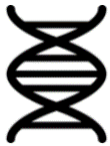


EB GENETICS

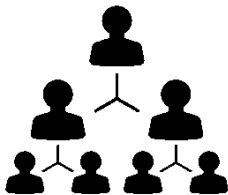
- Genes are the instructions that our cells use to make proteins.
- Each protein has a unique specialized job in the cell.
- Everyone has two copies of each of their genes (one from their mother and one from their father) – both parents contribute equally to the number of genes in their children.

EB genes



- 17 different genes that are known to cause the several different types of EB when they are defective.
- All these genes play a role in the formation of the epidermis (outer layer of the skin).
- When an EB gene stops working properly the protein it encodes is not made properly.

How is EB Inherited

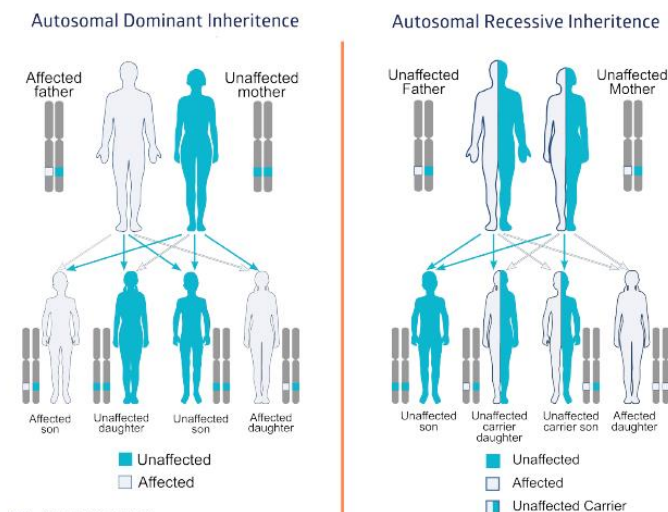


Autosomal Dominant

- Most forms of EB are autosomal dominant
- Mutation in only one of their EB gene
- Affect males and females equally
- Person with Dominant EB has a 50% chance of having a child with EB in each pregnancy

Autosomal Recessive

- Mutation in both copies of their EB gene
- Affect males and females equally
- Parents of affected children are “carriers” of a single mutation but do not have EB
- Two carrier parents have a 25% of have an affected child in each pregnancy



EB GENETICS

Genetic testing and counselling



Genetic testing may help families to:

- Identify the specific gene and mutation that caused EB,
- Pinpoint the exact type of EB that the patient has, and
- Help to identify other carriers of EB who would be at increased risk to have an affected child.

Genetic counselling may help families to:

- Better understand the EB diagnosis and how it is inherited,
- Identify who might be at risk to be a carrier or to be affected in the family,
- Explore the patient and family's feelings related to the diagnosis and connect them with appropriate resources, and
- Help the family understand their options for future family planning, including preimplantation genetic diagnosis (PGD) and prenatal diagnosis (PND).

Useful Websites/links:

- www.swrwoundcareprogram.ca
- EB Resources, including EB handbook: <https://www.debracanada.org>

Disclaimer: Unless stated otherwise, information contained in this document is taken from Epidermolysis Bullosa: A Handbook for EB Patients and Families, Developed by the Section of Dermatology at the Hospital for Sick Children with the Support of DEBRA Canada and Sick Kids Hospital Department of Dermatology EB Expert Team - all content used with permission. This document was created by SWRWCP (August, 2019)