EUROPEAN ACADEMY OF DERMATOLOGY & VENEREOLOGY

Information Leaflet for Patients

Inherited Epidermolysis Bullosa in Pregnancy

The aim of this leaflet

This leaflet has been written to help you understand more about inherited Epidermolysis Bullosa (EB), also called epidermolysis bullosa congenita and its management in and prior to pregnancy. It tells you what the condition is, what causes it, and how it can be treated

WWW.EADV.ORG
INFORMATION LEAFLET FOR PATIENTS 2024 2024 © EADV

Inherited Epidermolysis Bullosa in Pregnancy

What is Epidermolysis Bullosa?

Epidermolysis Bullosa is a group of genetic diseases characterized by very fragile skin. It manifests through the formation of blisters in response to seemingly minor or insignificant trauma or traction on the skin or mucous membranes. Depending on the type of genetic damage, blisters may form at different body sites, leading to surface wounds (with medical terms, erosions or ulcerations).

What causes Epidermolysis Bullosa?

Inherited Epidermolysis Bullosa is caused by unfavourable genetic changes (mutations) in the cells of the skin. These mutations mean the proteins, that hold the layers of the skin together, cannot work properly, causing the skin form blisters with minimal trauma.

How is Epidermolysis Bullosa diagnosed?

The diagnosis is established through a comprehensive examination of the newborn's or child's skin, searching for blisters, wounds or other signs. Additionally, a skin biopsy is performed to analyze the tissue and confirm the diagnosis. Lastly, genetic testing may be recommended to identify the specific genes responsible for EB.

Can Epidermolysis Bullosa be cured?

Currently, there is no general cure for Epidermolysis Bullosa. A few cases of severe, life-threatening forms have already been cured by gene therapy. However, this kind of treatment is not yet available and successful for everyone with EB.

How is Epidermolysis Bullosa treated?

At present, there are no targeted treatments available for most types of inherited Epidermolysis Bullosa. The day-to-day management focuses on preventing rubbing the skin, providing wound care, avoiding infections and pain management. In a few selected patients with severe disease, faulty genes can be replaced by corrected ones.

Does EB affect fertility?

In milder forms of EB, fertility is usually unaffected. However, in more severe forms of EB, associated with other medical conditions/problems (including poor nutrition and low body weight), the ability to ovulate (releasing an egg) or produce sperm may be affected. Severe type of EB can affect the moist soft tissue lining within vulva and vagina, called 'mucosa'. This mucosa is more fragile in EB, and can lead to pain during sexual intercourse and, in rare cases, become narrowed, developing a condition call 'vaginal stenosis'. Water-based lubricants use can be helpful.

Will my child inherit Epidermolysis Bullosa if I have it?

EB is a hereditary genetic disorder, indicating it can be passed down from parents to their offspring.

Please note that this information does not apply to autosomal recessive forms of EB (a rarer and much more severe form of EB). When one parent has EB, there's a 50% chance of their child inheriting the defective gene and consequently developing the condition. If both parents are carriers of the EB gene, even if they are asymptomatic, their child faces a 25% chance of inheriting the condition. It is advisable to undergo genetic testing and counselling if you are considering starting a family. If you are a carrier of one of the EB genes, genetic counselling should be performed prior to your pregnancy, and then prenatal diagnostics can be organized to prevent birth of newborns in case of life-threatening EB. Moreover, preimplantation genetic testing is also an option for in vitro fertilisation pregnancy plans.

Does Epidermolysis Bullosa worsen during pregnancy?

Many women experience morning sickness in pregnancy, as well as with acid reflux and symptoms of heartburn. This can be challenging for people with EB, who are at higher risk of already having long-term medical problems with the digestive system, as the common gestational symptoms exacerbate preexisting features of EB. Despite this, many women actually see improvements to their skin but may have a more challenging time with flare-ups post-partum. However, with careful monitoring and, if necessary, preventive treatment, it can be effectively managed.

Can women with EB have a vaginal delivery?

For women with any subtype of EB, vaginal birth should be recommended as the preferred delivery method. Notably, women with a severe form of EB called "recessive dystrophic epidermolysis bullosa" can undergo vaginal delivery without an increased risk of vaginal scarring.

For women expecting to have a baby with EB, vaginal delivery is still the recommended choice.

Can women with Epidermolysis Bullosa breastfeed?

Seeking guidance from a lactation consultant is strongly advised. Alternatively, you could reach out to your doctor or obstetrician. While some women with EB can breastfeed with problems, many can face challenges such as nipple blistering. This is not related to a specific subtype of EB but varies from person to person. Alternative options include breastfeeding with a shield, pumping, or supplementing with formula.

Where can I find more information about pregnancy with EB?

You can find more information about pregnancy, childbirth and aftercare in these internationally recognized guidelines: https://onlinelibrary.wiley.com/doi/full/10.1111/bjd.20809

Moreover, additional information concerning sexuality and EB is available at: https://www.debra-international.org/supporting-sexuality-in-eb-cpg.

EUROPEAN ACADEMY OF DERMATOLOGY & VENEREOLOGY

WWW.EADV.ORG
PATIENT LEAFLET 2024