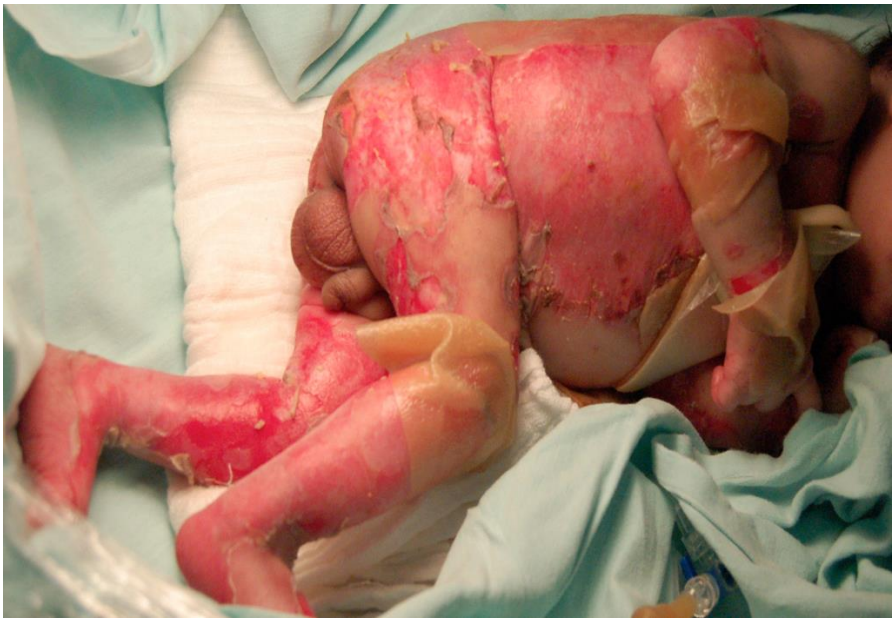
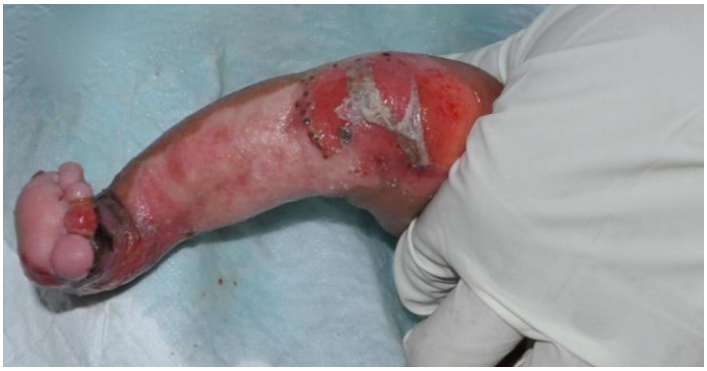
1. What Is Epidermolysis Bullosa (EB)?

EB is an umbrella term used to describe a group of inherited skin disorders that cause the skin to become very fragile.

In people with EB, the blisters may appear in response to minor trauma or friction from rubbing, scratching or adhesive tape. In severe cases, the blisters may occur spontaneously on skin & inside the body, such as the lining of the mouth or intestines.

Children with EB are known as **Butterfly children** because their skin is as fragile as the wings of a butterfly.





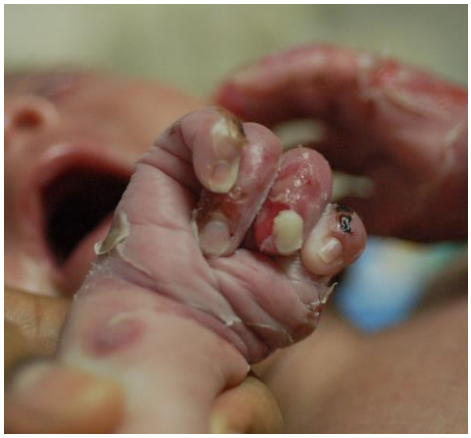


Fig 1.1 – 1.3: Blisters appear in response to minor trauma or friction from rubbing, scratching or adhesive tape

2. What Causes Epidermolysis Bullosa ?

The skin is made up of an outer layer (epidermis) and an underlying layer (dermis). The area where the layers meet is called the junctional zone. They are glued together by at least 15 types of skin protein (Collagen 7, Collagen 17, Keratin 5, Keratin 14, Plectin, etc)

Epidermolysis bullosa happens when one of the important skin protein is decreased or totally missing. The loss of this skin 'glue' makes the skin weak. Weakness of the protein is caused by a minor abnormality (a mutation) in the gene responsible for producing that protein.

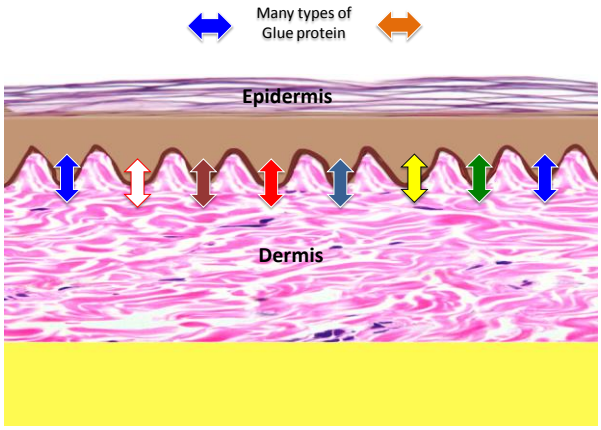


Fig 1: Normal skin structure

3. Is EB A Hereditary Disease?

Yes. It is an inherited disease.

EB is caused by faulty genes. In most cases these are inherited from one or both parents but sometimes the fault occurs spontaneously and appear for the first time in a person who has no other affected family member.

The inherited forms follow either autosomal dominant or autosomal recessive inheritance.

In autosomal recessive EB, neither parent has EB, but their child does. This happens when both parents are 'carriers' of the faulty genes. The risk for two carrier parents to both pass the defective gene and have an affected child is 25% with each pregnancy.

In autosomal dominant EB, only one abnormal gene is needed to express the disease. This means only one parent needs to carry the EB gene and the risk of passing the abnormal gene from affected parent to offspring is 50% for each pregnancy.

If you or your partner has EB or is known to be a carrier of a mutated gene associated with EB, it's possible to test an unborn baby at about 11 weeks into pregnancy.

4. How common is EB?

Based on international EB statistics, total number of people with EB worldwide is about 5 – 10 per million populations.

As our population is about 30 millions, it is estimated that there are about 150 – 300 Malaysians have EB in Malaysia. Currently, there are about 60 people with EB in our National EB registry. (DEBRA Malaysia).

5. What are the manifestations of EB?

The clinical manifestations of EB is based on the degree of skin fragility & associated internal organs involvement, the overall long term outcome ranges from mild to devastatingly severe.

The primary symptom of EB is blistering, which is often painful and which can sometimes significantly affect daily activities of living and interfere with school and work. EB usually occurs at birth or shortly after. EB affects both genders and every racial and ethnic background.

In milder form of EB, the manifestation of skin fragility (blisters) may not appear until a toddler first begins to walk or until an older child begins new physical activities that trigger more intense friction on the feet.

In more severe forms, there is generalized blistering of the skin due to trivial physical trauma or spontaneously without any apparent friction. Beside skin, blisters may also occur on internal organs, such as the oesophagus, stomach and respiratory tract.

These more severe forms of EB result in disfigurement, disability and early death, usually before the age of 30. In fact, some forms of EB are lethal in the first few months of life.

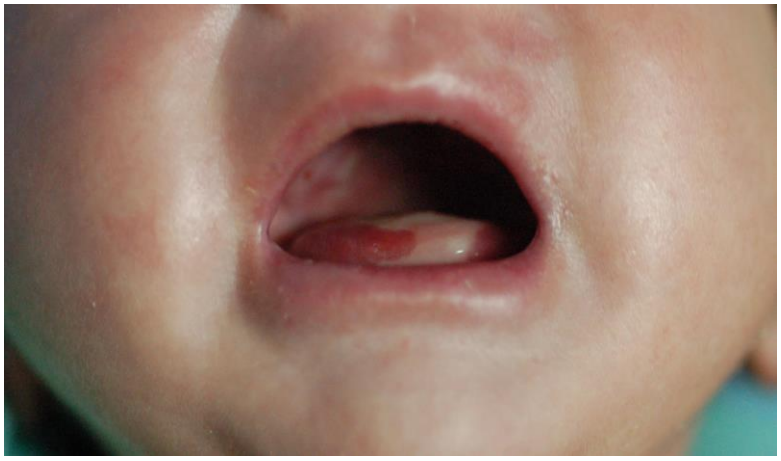
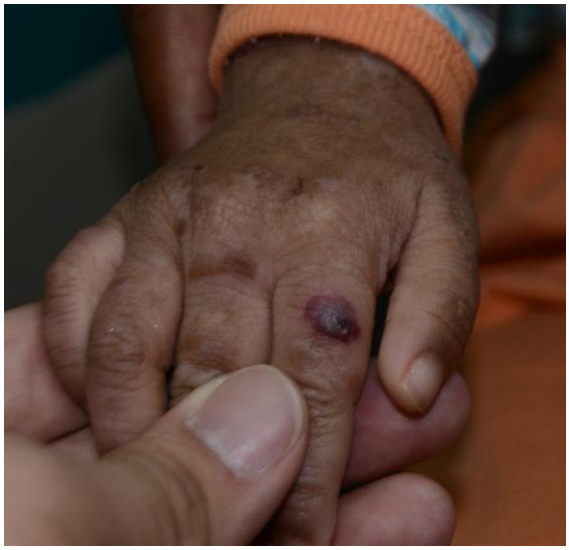


Fig 5.1: Blisters formation at mouth



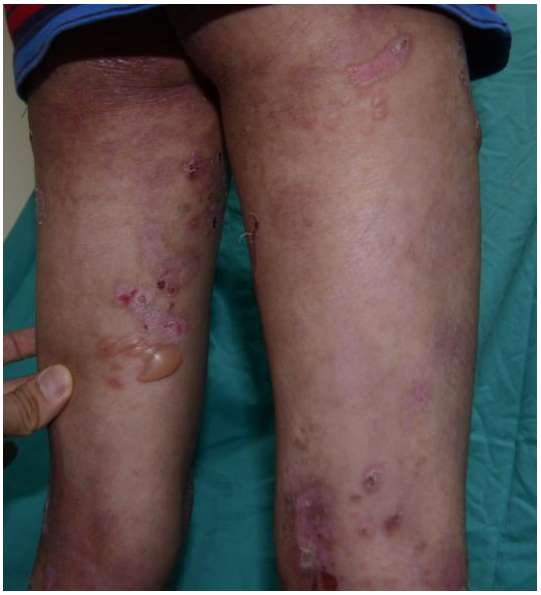


Fig 5.2 & 5.3: Blisters formation at hand & leg secondary to scratching

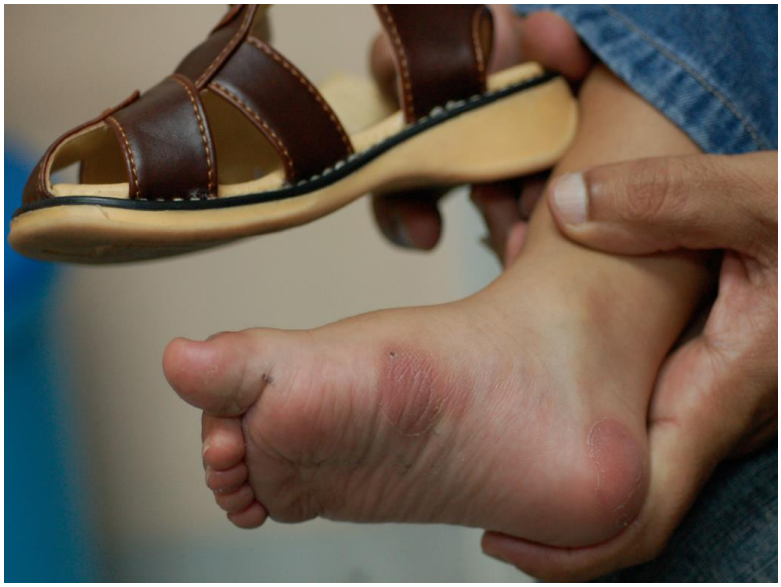


Fig 5.4: Blisters formation at feet secondary to friction

6. How will epidermolysis bullosa simplex be diagnosed?

The diagnosis of EB simplex can usually be based on the basis of the clinical history and the formation of the blisters that are related to trivial trauma. The diagnosis is further supported if other family members are affected as EB is an inherited disease.

In order to confirm the specific type of EB, your dermatologist may suggest taking a small sample of skin for more detailed microscopic examination with special staining. A blood test may be suggested to look for mutations of the genes likely to be involved.

Patients suspected of having EB can consult a qualified dermatologist for proper diagnosis and treatment. The names and place of practice of qualified dermatologists in Malaysia can be found on the Malaysian Association of Dermatologists (Persatuan Dermatology Malaysia) website – www.dermatology.org.my

7. How is EB treated?

EB is a lifelong disease for which there is currently no definitive treatment or cure, but the current researches and findings are promising.

Currently, treatment for severe forms of EB is focused on protecting the skin from trauma & blister formation, pain relief, promoting wound healing, preventing infection, attending to nutritional needs, and providing psychological support for the family.

A team of medical specialists from different disciplines (physicians, dietician, surgeon, dentist, psychologist, pain specialist and social/patient support group – DEBRA Malaysia) will help you decide what treatment is best for your child and offer advice about living with the condition.

Most treatments can be done at home by education and empowerment, such as popping blisters with a sterile needle, applying special non adhesive protective dressings and avoiding things that make the condition worse i.e. modify & reduce the friction related to the elasticated areas of a nappy or, during the crawling stage, the hands and knees etc

Medicines can be used to treat infection or to reduce pain.

Surgery can be used if EB causes narrowing of the food pipe or problems with the hands.

8. What is DEBRA Malaysia?

Persatuan Kebajikan Epidermolysis Bullosa Malaysia or Dystrophic Epidermoysis Bullosa Reasearch Association Malaysia (DEBRA Malaysia) is a charitable organization established in 2014 (Reg.No.PPM-001-14-10062014).

With dermatologists and representatives of EB family from different parts of Malaysia, DEBRA Malaysia aims to act as the body which will share information about EB management as well as psychosocial support to them.

For donation & further information,

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