

Assistance for professionals can be found at:

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Dermatologist
Auckland Dermatology
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Email: General: appt@dermatology.co.nz
Nick: nick.birchall@dermatology.co.nz
2. DEBRA NZ.
PO Box 7316
Wellington South
New Zealand
Telephone (& Fax): 04 389 7316
Email: trustees@debra.org.nz

Funded entirely from donations Debra NZ employs a part time specialist nurse to give assistance and advice to EB sufferers, their health practitioners, and their caregivers.

For new born babies urgent specialist advice is available for handling, feeding, woundcare and dressing techniques that can prevent unnecessary skin damage and minimise pain.
3. Online: www.debra.org.nz
Professional forums are also available at:
www.debra-international.org



Who is DebRA New Zealand?

DEBRA New Zealand has been running for twenty five years. It is a self help group for people and families with EB. A registered Charitable Trust DEBRA New Zealand relies on voluntary support and grants. New Zealand's Governor General Dame Sylvia Cartwright is the patron of DEBRA New Zealand.

DEBRA's primary objective is making life easier and more enjoyable for people and families who have to live with EB.

DEBRA New Zealand:

- Employs a part time specialist nurse to give assistance and advice to EB sufferers, their health practitioners, and their caregivers.
- Assists in collating the national register of EB patients and their subsequent carer support and wound dressings entitlements.
- Organises annual conferences and support networks for EB families.
- Organises life skills and adventure camps for Australasian teenagers and young adults with EB.
- Promotes EB awareness and education to the public, to health care professionals, and to government agencies.
- Raises money to fund EB support and research
- Contributes logistical support and expertise to EB organisations in other countries
- Supports and contributes to international research efforts

*Like a butterfly's wing EB sufferers are delicate and precious.
What they want most is to be like everyone else.*

DeBRA NEW ZEALAND
working for a life free of pain

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PO Box 7316 Wellington South New Zealand
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*Please note that all information given by DeBRA is for informational purposes only. Our information is not intended to substitute the care and guidance given by a qualified health professional. All regimens of care should be discussed with the patient's health professional. EB patients should always check with their health professional prior to starting any medications or treatment regimens.

What is EB?

A Clinician's Overview



What is EB?

Epidermolysis Bullosa (EB) is a rare genetic skin disorder which is characterised by skin fragility with blister formation occurring spontaneously or following minor trauma. This sometimes involves the mucous membranes.. EB can be broadly divided into three major categories that are recognised accordingly: **Simplex**, where cell lysis occurs in the epidermis; **Junctional**, where the separation occurs within the dermal-epidermal junction, and **Dystrophic**, in which the plane of cleavage is below the basement membrane in the dermis. These categories can be further sub-typed based on inheritance and clinical features. You cannot change from one type of EB to another.



EB ranges from mild to severe and can require major adjustments in the lifestyle of both the EB patient and his or her family. In severe EB, blisters are not confined to the outer skin. They may develop on the soft tissues (mucous membranes) inside the body such as the linings of the mouth, esophagus, stomach, intestines, lungs, airway, eyes and bladder. The extent of tissue involvement experienced by an individual is usually determined by the severity of the condition and the subtype present.

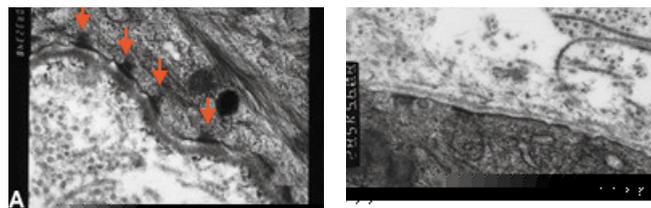
What causes EB?

A genetic change or mutation, in one of the genes that code for the proteins that "glue" the skin together. The particular protein affected is then reduced or missing in a specific layer of the skin, causing areas of structural weakness. As a result, the fragile skin is vulnerable to damage from mild friction, causing the blisters, which are the characteristic feature of EB. Sometimes these same proteins are also important for "glueing" internal tissues together, and hence in these cases, internal blistering/damage can result as well.

Diagnosis of EB

Diagnosis of EB is first dependant on the patient's clinical presentation and family history. Information from electron microscopy and immunofluorescence mapping from a skin biopsy is required to make the diagnosis.

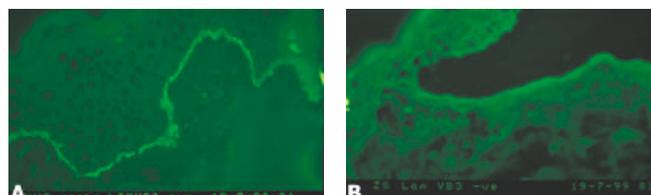
Electron microscopy involves the use of a highly powered microscope that evaluates the skin sample by looking at the level of skin separation present and also noting the number



Electron micrographs of the dermal-epidermal junction of skin samples obtained by a skin biopsy and found to be normal (A) and affected with JEB (B). The hemidesmosomes (arrows) are formed in normal skin and absent in the affected skin. Source: Associate Prof. Dédée F. Murrell, Head of Dermatology St George Hospital University of NSW, Sydney.

and appearance of specific structures, such as anchoring fibrils or hemidesmosomes, located within the skin. This will assist in diagnosing the correct subtype of EB.

Immunofluorescence mapping involves using a fresh skin sample and a panel of antibodies tagged with a fluorescent marker to detect the presence, or degree of binding, of a panel of antibodies to the skin proteins affected in EB. This may provide an indication as to the specific protein involved in the structural weakness of the skin. To refer a patient for this test, your doctor can have forms and transport media sent out by contacting Prof Dede Murrell, Head of Dept of Dermatology, St George Hospital, Kogarah, Sydney on + 61-2-9350-2543.



Immunofluorescence mapping of normal skin (A) and affected (B). Source: Associate Prof. Dédée F. Murrell, Head of Dermatology St George Hospital University of NSW, Sydney.

Waiting for these results can be difficult, so it is helpful to locate measures of support. Support systems such as family, other parents with children affected by EB and organisations such as DebRA (Dystrophic epidermolysis bullosa Research Association) can help during this trying time by providing support and information to families of affected individuals.

How is EB Inherited?

Autosomal Dominant Inheritance: An autosomal dominant disorder is one in which one gene for the condition expresses itself in an individual. A parent with an autosomal dominant form of EB has a 50:50 chance with each pregnancy of transmitting the abnormal gene. The chance is the same whether the child is a boy or a girl, and birth order does not make a difference. A child who does not inherit the gene for EB from an

affected parent will not have the condition and cannot pass it on.

Autosomal Recessive Inheritance: An autosomal recessive disorder is one in which a recessive (unexpressed) gene for the disorder is passed from each parent and the two genes are paired together, causing the disorder to be expressed in the child. If a person has one recessive EB gene paired with a normal gene, the person is "a carrier", but does not have the disorder. If parents are each carriers of an autosomal recessive gene, there is a 25 percent chance with each pregnancy that their children will have the disorder. Again, the sex of the child and the birth order do not matter. 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.

Once the genetic mutation is identified in a family, prenatal diagnosis of future pregnancies maybe possible.

Who gets EB?

Just over one hundred New Zealanders have EB. Nine children and five adults have a severe form of the disorder. It is estimated that there are around 1,000 people in Australia who have some form of EB and over 500,000 worldwide. It occurs in all racial and ethnic groups and affects males and females equally. EB may not always be evident at birth. Milder cases of EB may become apparent when a child crawls, walks, runs or when young adults become more physically active.

How is EB Treated?

Because EB involves many parts of the body, parents and health professionals must take a multi disciplinary approach to the treatment of an EB patient. Total patient care often must be provided, particularly for young children. The severe forms of EB require meticulous nursing care in regards to dressing changes and wound care. Much of this care is often provided by the parents; however, the education of all people who have contact with the patient is essential, including all health professionals, as well as teachers, relatives and others.

Although there is no cure for EB, and gene therapy is still in the realm of experimental medicine treatment of EB is directed towards the symptoms. Many persons with milder forms have minimal symptoms and may require less or no treatment. Management of this disorder is usually under the care of a Dermatologist as well as many other specialists as required. Focus should be on prevention of infection, protection of the skin against trauma, attention to nutritional deficiencies and dietary complications, minimisation of deformities and contractures, and the need for support to the entire family.