

## **Spontaneous healing of the hereditary skin disease EB is not rare**

**Spontaneous healing of the hereditary skin disease epidermolysis bullosa (EB) is more common than was generally thought. This conclusion is based on research of the department of Dermatology of the University Medical Center Groningen, Netherlands, led by Prof. Marcel Jonkman. A publication appeared in the New England Journal of Medicine (April 16, 2009).**

For patients suffering from EB, the most simple daily activities can cause the formation of painful blisters, due to mutation in genes that produce proteins that ensure that the various layers of the skin adhere together. Children with this disease are also known as Butterfly Children due to their skin being as fragile as the wings of a butterfly. Jonkman and Pasmooij found that 7 of the 20 patients (35%) with EB had several skin patches, which showed spontaneous healing. The patches could be recognized by their homogenous pigmentation and the lack of blisters.

### **Cause of spontaneous healing**

The phenomenon of spontaneous healing, also called revertant mosaicism, was described for the first time in 1997 by the same research group from Groningen. The underlying cause was a spontaneous back mutation of the inherited disease-causing gene in the skin cells. Now, their research showed that these back mutations are more common than was expected, and that various of these patches can appear in a patient with EB. Every patch was repaired by another back mutation.

### **Skin transplant**

The research in the UMCG aims to use the patient's healthy skin cells to heal the affected skin. This treatment of the hereditary disease EB is called revertant cell therapy. In revertant cell therapy, healthy skin cells are stimulated to grow in the laboratory in order to develop a skin transplant. The transplant is subsequently used to substitute the diseased skin. The high percentage of revertant mosaicism means that more patients could benefit from this new treatment. The UMCG investigates the results of this treatment in cooperation with a research group in Madrid. The preparations for the skin transplant in the UMCG are currently ongoing.

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