BIOENGINEERED SKIN FOR THE TREATMENT OF RECESSIVE DYSTROPHIC EPIDERMOLYSIS BULLOSA

INNOVATION AND DESCRIPTION OF THE TECHNOLOGY

Recessive dystrophic epidermolysis bullosa (RDEB) is a subtype of DEB characterised by generalized cutaneous and mucosal blistering. Specifically, RDEB is a severe skin fragility genodermatosis caused by loss-of-function mutations in the COL7A1 gene, encoding type VII collagen (C7). C7 deficiency results in generalized blistering of the skin and other stratified epithelia, scarring, fibrosis, mitten-like deformities of hands and feet, and a high risk of developing metastatic squamous cell carcinoma.

The present medicinal product discloses an autologous bioengineered skin containing polyclonal populations of RDEB patient keratinocytes and fibroblasts corrected by a non-viral gene editing strategy.

The technology is based on genetic correction of keratinocytes and fibroblasts will allow expression of type VII collagen, C7, by both cell types leading, in turn, to anchoring fibril formation and restoration of dermal-epidermal adhesion. The use of a bioengineered skin containing fibroblasts and keratinocytes represents an advantage over single sheets of genetically corrected epidermal cells in terms of handling (mechanical resistance) and resemblance of the native skin situation where both cell types produce C7.

MARKET AND ADVANTAGES OF THE TECHNOLOGY

The field of application of the orphan drug is patients with RDEB who carry pathogenic mutations in exon 80 of the COL7A1 gene. In Spain, 46 % of the EBDR alleles are carriers of the c.6527insC mutation. But also this technology could be useful for others exons of this gene, being able to be effective for a large number of RDEB patients

Bioengineered skin containing CRISPR/Cas9 gene-edited keratinocytes through a non-viral precise excision of exons encoding COL7A1 collagenous domain, is not authorized anywhere worldwide for the treatment of RDEB.

The innovation of the present technology is based on the use of an accuracy gene-correction method (edition instead of addition) of extremely high efficiency that allows the immediate application of corrected cells without selection of edited cell clones.

Furthermore, another key point is the use of a non-viral gene transfer system instead of using a viral vector which has potential risks associated with their genomic integration.

IPRS AND CONTACT

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