

NEW THERAPY FOR THE TREATMENT OF EPIDERMOLYSIS BULLOSA

Results achieved from researches from CIBER, CIEMAT, UC3M and IIS-FJD offer an evidence of an ex vivo effective genome editing tool able to achieve precise gene correction in primary RDEB keratinocytes as new treatment for EB.

The necessity

Epidermolysis Bullosa refers to a group of diseases characterized by strong skin fragility. The recessive dystrophic subtype (RDEB) is the most severe phenotype of the disease.

A large number of mutations have been described within *COL7A1* gene, making this gene the main target of gene therapy strategies RDEB.

The solution

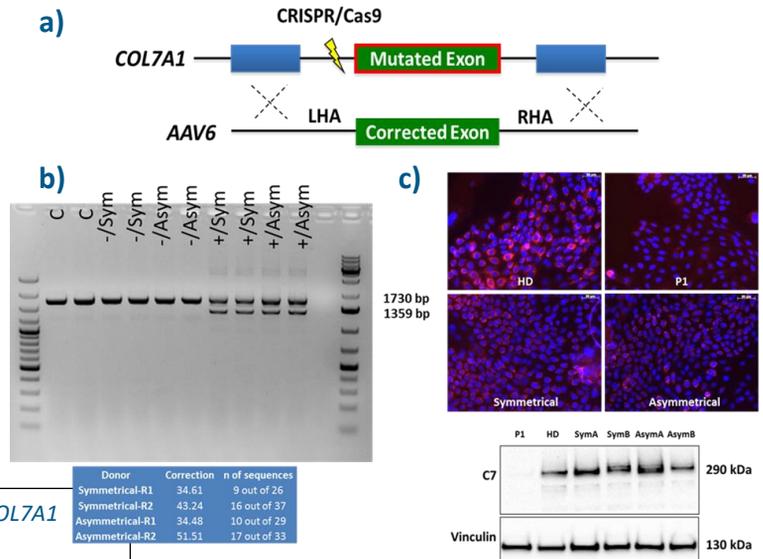
Recently, benefits have been shown in an ex vivo phase I clinical trial in a patient transplanted with skin equivalents, but new gene editing tools open the door to new advanced therapies.

The present invention disclose the most efficient tool for genome editing in primary keratinocytes (a cell type considered hard-to-transfect).

Innovative aspects

For the first time, a large collection of AAV serotypes have been tested in order to find the highest transduction efficiency in primary keratinocytes. Also, it is possible to develop a large collection of AAVs containing different regions of *COL7A1*, correcting any mutation along the gene.

Furthermore, the present technology is capable of achieve close to 40% of precise gene correction in primary keratinocytes, surpassing previous HR-bases gene correction ratios.



- a) Scheme of the HDR-based strategy for precise *COL7A1* correction.
- b) HDR-based correction genotyping in primary keratinocytes.
- c) Immunofluorescence analysis and western blot analysis.

Intellectual Property

Priority European patent application filed (January 20, 2020)
Suitable for international extension (PCT application)

Stage of Development:

Developing pre-clinical studies required to apply for a Phase I/II clinical trial

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