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| Título del Proyecto | **Mutation-targeted gene and pharmacological therapies for dystrophic and junctional Epidermolysis Bullosa** |
| Nº de expediente asignado | AC17/00054 |
| Abstract | The goal of translating new experimental therapies for rare diseases to the clinic remains a formidable challenge. Inherited skin fragility disorders such as Epidermolysis Bullosa (EB) are currently the target of promising therapeutic approaches, some already in clinical trials.<br>While standard ex vivo gene therapy strategies for different forms of EB are valid, they remain costly and are subject to tough health regulatory policies. They also pose potential genotoxic risks. Therefore, new, innovative, safer and efficacious approaches to restore expression of adhesive skin proteins in EB patients should be explored. With this premise in mind and supported by strong preliminary data, we propose in this project several potential therapies to correct recurrent mutations causing recessive dystrophic EB and junctional EB that will be advanced through in vitro testing and assessed in vivo using surrogate skinhumanized mouse models that faithfully recapitulate the human EB skin conditions. The therapies are: i) nuclease-facilitated, NHEJ-driven gene edition to remove and replace mutations, ii) antisense oligonucleotides enabling splicing modulation (skipping) of mutant exons and iii) new drugs promoting the readthrough of nonsense mutations. The MuTaEB consortium gathers together outstanding research and clinical groups with complementary expertise to jointly attack the disease from several fronts. Together we hope to bring new therapies to the clinic. |
| Entidad Financiadora | Instituto de Salud Carlos III (ISCIII) |
| Convocatoria: | E-Rare-3 “Transnational Research Projects for Innovative Therapeutic Approaches for Rare Diseases”.<br>ACCIONES COMPLEMENTARIAS DE PROGRAMACION CONJUNTA INTERNACIONAL – AES 2017. |

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| Importe de la ayuda | 130.680€ |
| Fechas de ejecución del proyecto | 01/01/2018-31/12/2020 |
| | Estas ayudas están financiadas por el Instituto de Salud Carlos III y cofinanciadas por FEDER, Programa Operativo Crecimiento Inteligente 2014-2020 “Una manera de hacer Europa” |
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| Enlaces: | http://www.erare.eu/financed-projects/mutaeb<br><br>https://www.ciberisciii.es/areas-tematicas/grupo-de-investigacion?id=17093 |