TITLE: PKD GENE EDITING TREATMENT METHOD

FIELD OF INTEREST

Biotechnology (Gene Editing, Pyruvate kinase deficiency)

CLINICAL NEED

Pyruvate kinase deficiency (PKD) is the most common erythroid inherited enzymatic defect causing chronic non-spherocytic hemolytic anemia. PKD is an autosomal recessive disorder caused by mutations in the PKLR gene. This gene encodes for two different transcript variants, RPK and LPK, expressed in red blood cells and liver respectively. To date, more than 200 different mutations in the PKLR gene have been linked to PKD.

Over the last few years, gene editing has emerged as a promising gene therapy approach for blood-cell disorders, since genetic mutations can be accurately corrected. One of the current bottlenecks to use gene editing to treat inherited disorders is the generation of DSB in places different from the selected specific on-target site that can generate undesired genetic modifications with unexpected clinical effects.

DESCRIPTION OF THE INVENTION

Researchers propose a new treatment of Pyruvate kinase deficiency (PKD) using gene therapy. This new technology offers the possibility to minimize the potentially harmful off-target mutations, which are the main problem of the gene edition therapies.

TECHNOLOGY KEYWORDS

PKD, Gene Editing, PKLR gene.

IPR STATUS

Patent application number: EP20382568.

Applicants: CIEMAT, CIBER, FIIS-FJD and UNIV. STANFORD.

TYPE AND ROLE OF PARTNER

Looking for commercial partners interested in licensing.

Contact details:

Innovation Unit
e-mail: irene.ruano@fjd.es; laura.felipeo@quironsalud.es