GENE EDITING AS A TREATMENT OF PYRUVATE KINASE DEFICIENCY (PKD)

The present Technology developed by researchers from CIBER, CIEMAT and FIIS-FJD provides a method for the treatment of PKD using CRISPR system

The Need

PKD is an autosomal recessive disorder caused by mutations in the *PKLR* gene. PKD is the most common erythroid inherited enzymatic defect causing chronic non-spherocytic hemolytic anemia.

The prevalence of PKD is estimated at 1.9 cases per 100,000 people in the Caucasian population.

The Solution

Gene editing has emerged as a promising gene therapy approach for blood-cell disorders, since genetic mutations can be accurately corrected.

The clinical use of gene editing therapy to correct PKD is likely. However, the most concerning issue of this new gene editing technology to be applied to he clinic is the off-target effect caused by the ribonucleoprotein complex (RNPs).

Innovative Aspects

The present technology avoid off-target effects. It is possible due to the location of the double-strand break (DSB) as closely as possible to the place where the exogenous DNA is going to be integrated.

Based on research results, the invention relates the use of a crRNA to provide a novel RNP complex for use in gene editing therapy to correct PKD in order to eliminate or reduce the off-target effects caused by the RNP.

On the whole, the present invention offers evidence of a RNP complex which, together with the use of Adeno-associated virus (AAV) is especially suitable as an ex vivo effective genome editing tool.

Intellectual Property

- Priority European patent application filed
- Suitable for international extension (PCT application)

Aims

Looking for a partner interested in a license and/or collaboration agreement to develop and exploit this asset

Contact details

Centro de Investigación Biomédica en Red *(CIBER)*
gemma.gomez@ciberisciii.es
transferencia@ciberisciii.es
https://www.ciberisciii.es/en