# *EX VIVO GENE THERAPY APPROACH IN DIAMOND BLACKFAN ANEMIA (DBA) WITH LENTIVIRAL VECTORS*

**Preclinical studies developed by the Division of Hematopoietic Innovative Therapies from CIBER-CIEMAT-FIIS demonstrate the efficacy and the safety of an ex vivo gene therapy approach in DBA with lentiviral vectors**

## The Need

DBA is a rare disease with an estimated prevalence of 5-10 per million life births. The hallmark of DBA is macrocytic anemia, typically presented at the first year, and frequently evolves to neutropenia and thrombocytopenia, and in some cases to a myelodisplac syndrome or acute myeloid leukemia. Mutations in 20 DBA genes, plus 3 "DBA–like" genes account for 70-80% of DBA patients.

## The Solution

The technology is based on that, the ex vivo gene correction of HSPCs from DBA patients may also confer a proliferative advantage on their HSPCs, thus favouring the progressive improvement of the haematological status of these patients. Therefore, this proposal was focused on the development of the preclinical studies that would be required to develop a subsequent gene therapy trial in DBA patients with mutations in RPS19.

## Innovative Aspects

According to the expertise of our group and collaborators in the biology, clinical management of patients and also on the gene therapy of another severe BMF syndrome and another rare anemia such as pyruvate kinase deficiency (PKD), the group are in an ideal position to be the first one to develop worldwide a new, efficient and safe cure for patients with DBA.

The group consider the possibility of conducting the clinical trial in unconditioned patients, implying a minimal risk associated to the gene therapy of DBA patients. In this way, this project will contribute to the development of a potential cure for DBA and the whole project will be carried out with the close collaboration of DBA patient associations.

The project has obtained funding from the EJP on Rase Diseases

**Stage of Development:**
Ending the pre-clinical phase in 2022. Phase I/II clinical trial in 2023

## Intellectual Property

- PCT application filed
- ODD application filed

## Aims

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

## Contact details

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