RARE DISEASE

Novel treatment to mitigate the Ectrodactyly-Ectodermal dysplasia-Clefting (EEC) associated blindness

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Protection	IT 102020000023647 EP 21799330.2
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What it is needed for?

The **Ectrodactyly-Ectodermal dysplasia-Clefting (EEC) syndrome** is a rare genetic disorder due to mutations in the p63 gene and characterized by limb defects, orofacial clefting, ectodermal dysplasia, and **ocular surface defects that lead to the loss of vision**.

Currently **no** specific **treatments** for this pathology **exist** leaving patients with supportive care to alleviate symptoms and thus greatly reduced quality of life.

We have identified a novel therapeutic **strategy to preserve** patient's – affected by EEC syndrome– **vision**. We have demonstrated that applying a **proprietary siRNA** contrasts untoward effects of a causal mutation in the p63 gene in the patient derived models of the disease.

This finding provides a basis for the development of a **therapeutic** strategy that **targets the cause of the disease**.

Advantages

- No specialized treatment for ECC syndrome exists. We propose the solution that targets the cause of the disease.
- A treatment is targeted: it affects only a mutant, but not the WT allele.

Applications

A therapeutic for the mitigation of EEC associate corneal blindness, and/or other disorders/alterations due to the R279H mutation in the p63 gene.

What we are looking for

Technology is available for licensing and/or co-development

TRL scale

Discovery | **Lead Optimization** | **Preclinical** | **Clinical Phases**

Health and Biomedical - Pharmaceuticals