Group name

Case name
Therapy of type 2 autosomal dominant osteopetrosis (ADO2) caused by mutation of the CLCN7 gene (ADO2 CLCN-dependent).

Owner
University of L'Aquila

Website
http://www.univaq.it

description

The invention is in the field of molecules known as "small interfering RNA" with therapeutic applications. These molecules have the ability to reduce the expression of genes in an extremely specic way, allowing the function of cells, thanks to molecular manipulations previously precluded. The CLCN7- dependent ADO is a disease characterized by the absence of bone cell function called osteoclasts, and manifests itself with very dense but brittle bones, disorders in the production of blood cellular elements and sensory-motor functions, inflammation of the osteo-muscular apparatus, and dental problems. The disease, currently untreated, affects 5 individuals out of 100,000, and is caused by the mutation of a gene (CLCN7), essential for the function of osteoclasts.

In this context, a team of researchers from the University of L'Aquila developed siRNAs optimized for the treatment of ADO2 CLCN7-dependent disease, which act on the selective reduction of the mutated protein compared to the normal one.

The siRNA molecules developed have proved to be highly specic, **eliminating at least 80% of the transcript of the mutated gene**, restoring the function of the osteoclasts, and thus creating a situation similar to the normal functioning of the cells, and an improvement in the symptoms of the disease.

Date
2019-01-29 00:00:00

Case Ref.
2

Contact person
Luigi Di Domenico

Other
Alessandra Montesanti

Link to EPO database
https://worldwide.espacenet.com/publicationDetails/description?CC=EP&NR=3145553A1&KC=A1&FT=D&ND=4&date=20170329&DB=&locale=en_EP#

Industry
Pharmaceutical

Transaction type
Subject to negotiation

Application number
WO2015IB53730 20150521

Applicants
UNIVERSITA' DEGLI STUDI DELL'AQUILA

Inventors
TETI ANNA MARIA; RUCCI NADIA; CAPULLI MATTIA; MAURIZI ANTONIO

Limitations:

Pre-clinical development phase of therapy for ADO2 and characterization on a mouse model (animal).

Meta information:

Meta title
Therapy of type 2 autosomal dominant osteopetrosis (ADO2) caused by mutation of the CLCN7 gene (ADO2 CLCN-dependent).

Meta description

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Support:

Access to additional documentation
Please inquire

Support from inventors
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Geographic scope:

Regions
PCT

Classification

IPC classifications
119934

119935

CPC classifications
119936

119937

119938

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