

HORIZON
2020

Repurposing of carbamazepine for treatment of skeletal dysplasia

Fact Sheet

Project Information

MCDS-Therapy

Grant agreement ID: 754825

[Project website](#) 

DOI

[10.3030/754825](https://doi.org/10.3030/754825) 

Project closed

EC signature date

5 December 2017

Start date

1 December 2017

End date

31 May 2024

Funded under

SOCIETAL CHALLENGES - Health, demographic change and well-being

Total cost

€ 5 697 390,50

EU contribution

€ 5 697 390,50

Coordinated by

UNIVERSITY OF NEWCASTLE
UPON TYNE United Kingdom

Objective

Genetic skeletal diseases (GSDs) are an extremely diverse and complex group of rare genetic diseases that affect the development the skeleton. There are more than 450 unique and well-characterised phenotypes that range in severity from relatively mild to severe and lethal forms. Although individually rare, as a group of related genetic skeletal diseases, GSDs have an overall prevalence of at least 1 per 4,000 children, which extrapolates to a minimum of 225,000 people in the 27 member states and candidate countries of the EU. This burden in pain and disability leads to poor quality of life and high healthcare costs.

Metaphyseal chondrodysplasia, type Schmid (MCDS) results from mutations in

collagen X and affects <1/100,000 of the population. Mutant collagen X molecules miss-fold during synthesis and are retained within the endoplasmic reticulum (ER) of hypertrophic chondrocytes, thereby causing ER stress. Our extensive pre-clinical studies have shown that carbamazepine (CBZ) can alleviate ER stress caused by the expression of mutant collagen X and restore bone growth in a validated mouse model of MCDS. CBZ is an FDA approved drug used for the treatment of epilepsy and bipolar disorder and received orphan drug designation by the European Commission for the treatment of MCDS in September 2016.

MCDS-Therapy was originally proposed as a 5-year collaborative project comprising world-renown clinical centres and SMEs to advance the repurposing of CBZ for MCDS (up to the Marketing Authorization Application dossier) through a multicentre and multinational (EU & AUS) clinical trial (Phase1, Phase2/3). MCDS-Therapy also encompasses biomarker development and health economics assessment studies to deliver, evidence to inform potential further studies of an innovative and affordable (CBZ already exists in a generic form) repurposed therapy for MCDS along with the diagnosis/prognosis tools to personalise the treatment strategy. The original proposal was for completion of this by 2022 however delays associated with the COVID pandemic have resulted in a need to extend the project with completion now forecast by May 2024.

Fields of science (EuroSciVoc)

[medical and health sciences](#) > [basic medicine](#) > [neurology](#) > **[epilepsy](#)**

[medical and health sciences](#) > [health sciences](#) > [public health](#) > [epidemiology](#) > **[pandemics](#)**

[natural sciences](#) > [biological sciences](#) > [genetics](#) > **[mutation](#)**



Keywords

[Orphan drug](#)

[Repurposing](#)

[clinical trial](#)

[Health economics](#)

[biomarkers](#)

[skeletal dysplasia](#)

[metaphyseal chondrodysplasia type Schmid](#)

Programme(s)

[H2020-EU.3.1. - SOCIETAL CHALLENGES - Health, demographic change and well-being](#)

MAIN PROGRAMME

[H2020-EU.3.1.3. - Treating and managing disease](#)

Topic(s)

[SC1-PM-08-2017 - New therapies for rare diseases](#)

Call for proposal

[H2020-SC1-2016-2017](#) 

[See other projects for this call](#)

Sub call

H2020-SC1-2017-Two-Stage-RTD

Funding Scheme

[RIA - Research and Innovation action](#)

Coordinator



UNIVERSITY OF NEWCASTLE UPON TYNE

Net EU contribution

€ 1 960 601,86

Total cost

€ 2 370 889,12

Address

KINGS GATE

NE1 7RU Newcastle Upon Tyne

 **United Kingdom** 

Region

North East (England) > Northumberland and Tyne and Wear > Tyneside

Activity type

Higher or Secondary Education Establishments

Links

[Contact the organisation](#)  [Website](#) 

[Participation in EU R&I programmes](#) 

[HORIZON collaboration network](#) 

Participants (11)

 THIRD-PARTY 

THE NEWCASTLE UPON TYNE HOSPITALS NHS FOUNDATION TRUST

 United Kingdom

Net EU contribution

€ 410 287,26

Address

FREEMAN HOSPITAL, HIGH HEATON
NE7 7DN Newcastle-Upon-Tyne 

Region

North East (England) > Northumberland and Tyne and Wear > Tyneside

Activity type

Public bodies (excluding Research Organisations and Secondary or Higher Education Establishments)

Links

- [Contact the organisation](#) 
- [Participation in EU R&I programmes](#) 
- [HORIZON collaboration network](#) 

Total cost

No data



ASSISTANCE PUBLIQUE HOPITAUX DE PARIS

 France

Net EU contribution

€ 99 677,72

Address

55 BOULEVARD DIDEROT
75012 Paris 

Region

Ile-de-France > Ile-de-France > Paris

Activity type

Research Organisations

Links

- [Contact the organisation](#) 
- [Website](#) 

[Participation in EU R&I programmes](#)
[HORIZON collaboration network](#)

Total cost
€ 99 677,72



UNIVERSITAIR ZIEKENHUIS ANTWERPEN

Belgium

Net EU contribution
€ 70 376,53

Address
DRIE EIKENSTRAAT 655
2650 Edegem

Region
Vlaams Gewest > Prov. Antwerpen > Arr. Antwerpen

Activity type
Research Organisations

Links
[Contact the organisation](#) [Website](#)
[Participation in EU R&I programmes](#)
[HORIZON collaboration network](#)

Total cost
€ 70 376,53



UNIVERSITAETSKLINIKUM FREIBURG

Germany

Net EU contribution
€ 382 015,88

Address
HUGSTETTER STRASSE 49
79106 Freiburg

Region
Baden-Württemberg > Freiburg > Freiburg im Breisgau, Stadtkreis

Activity type
Higher or Secondary Education Establishments

Links

- [Contact the organisation](#)
- [Website](#)
- [Participation in EU R&I programmes](#)
- [HORIZON collaboration network](#)

Total cost

€ 382 015,88



MURDOCH CHILDRENS RESEARCH INSTITUTE

Australia

Net EU contribution

€ 340 625,00

Address

FLEMINGTON ROAD RCH
3052 Parkville

Activity type

Research Organisations

Links

- [Contact the organisation](#)
- [Website](#)
- [Participation in EU R&I programmes](#)
- [HORIZON collaboration network](#)

Total cost

€ 340 625,00



GUYS AND ST THOMAS' NHS FOUNDATIONTRUST

United Kingdom

Net EU contribution

€ 277 428,75

Address

Westminster Bridge Road
SE1 7EH London

Region

London > Inner London — East > Lambeth

Activity type

Research Organisations

Links

- [Contact the organisation](#)
- [Website](#)
- [Participation in EU R&I programmes](#)
- [HORIZON collaboration network](#)

Total cost

€ 277 428,75



BEACON: FOR RARE DISEASES LTD

United Kingdom

Net EU contribution

€ 309 235,00

Address

66 DEVONSHIRE ROAD
CB1 2BL Cambridge

Region

East of England > East Anglia > Cambridgeshire CC

Activity type

Research Organisations

Links

- [Contact the organisation](#)
- [Participation in EU R&I programmes](#)
- [HORIZON collaboration network](#)

Total cost

€ 309 235,00



ISTITUTO ORTOPEDICO RIZZOLI

Italy

Net EU contribution

€ 571 741,84

Address

VIA DI BARBIANO 1/10
40136 Bologna

Region

Nord-Est > Emilia-Romagna > Bologna

Activity type

Research Organisations

Links

[Contact the organisation](#)  [Website](#) 

[Participation in EU R&I programmes](#) 

[HORIZON collaboration network](#) 

Total cost

€ 857 175,00

 **THIRD-PARTY** 

ALMA MATER STUDIORUM - UNIVERSITA DI BOLOGNA

 Italy

Net EU contribution

€ 285 433,16

Address

VIA ZAMBONI 33

40126 Bologna 

Region

Nord-Est > Emilia-Romagna > Bologna

Activity type

Higher or Secondary Education Establishments

Links

[Contact the organisation](#)  [Website](#) 

[Participation in EU R&I programmes](#) 

[HORIZON collaboration network](#) 

Total cost

No data



SCiOMICS GMBH

 Germany

Net EU contribution

€ 604 030,00

Address

KARL-LANDSTEINER STR. 6

69151 Neckargemund 

SME 

Yes

Region

Baden-Württemberg > Karlsruhe > Rhein-Neckar-Kreis

Activity type

Private for-profit entities (excluding Higher or Secondary Education Establishments)

Links

[Contact the organisation](#) 

[Participation in EU R&I programmes](#) 

[HORIZON collaboration network](#) 

Total cost

€ 604 030,00



FINOVATIS

 France

Net EU contribution

€ 385 937,50

Address

62 RUE DE BONNEL

69003 Lyon 

SME 

Yes

Region

Auvergne-Rhône-Alpes > Rhône-Alpes > Rhône

Activity type

Private for-profit entities (excluding Higher or Secondary Education Establishments)

Links

[Contact the organisation](#)  [Website](#) 

[Participation in EU R&I programmes](#) 

[HORIZON collaboration network](#) 

Total cost

€ 385 937,50

Last update: 19 February 2025

Permalink: <https://cordis.europa.eu/project/id/754825>

European Union, 2025

