

# Integrated European –omics research project for diagnosis and therapy in rare neuromuscular and neurodegenerative diseases

<https://neurodegenerationresearch.eu/survey/integrated-european-%c2%96omics-research-project-for-diagnosis-and-therapy-in-rare-neuromuscular-and-neurodegenerative-diseases/>

## **Principal Investigators**

### **Institution**

### **Contact information of lead PI**

### **Country**

European Commission

## **Title of project or programme**

Integrated European –omics research project for diagnosis and therapy in rare neuromuscular and neurodegenerative diseases

## **Source of funding information**

European Commission FP7-Seventh Framework Programme

## **Total sum awarded (Euro)**

€ 12,000,000

## **Start date of award**

01/10/2012

## **Total duration of award in years**

5.0

## **The project/programme is most relevant to:**

Neurodegenerative disease in general

## **Keywords**

### **Research Abstract**

Neurodegenerative (ND) and neuromuscular (NM) disease is one of the most frequent classes of rare diseases, affecting life and mobility of 500,000 patients in Europe and millions of their caregivers, family members and employers. This NEUROMICS project brings together the leading research groups in Europe, five highly innovative SMEs and relevant overseas experts using the most sophisticated Omics technologies to revolutionize diagnostics and to develop

pathomechanism-based treatment for ten major ND and NM diseases. Specifically we aim to:

- (i) use next generation WES to increase the number of known gene loci for the most heterogeneous disease groups from about 50% to 80%,
- (ii) increase patient cohorts by large scale genotyping by enriched gene variant panels and NGS of so far unclassified patients and subsequent phenotyping,
- (iii) develop biomarkers for clinical application with a strong emphasis on presymptomatic utility and cohort stratification,
- (iv) combine -omics approaches to better understand pathophysiology and identify therapeutic targets,
- (v) identify disease modifiers in disease subgroups cohorts with extreme age of onset
- (vi) develop targeted therapies (to groups or personalized) using antisense oligos and histone deacetylase inhibitors, translating the consortiums expertise in clinical development from ongoing trials toward other disease groups, notably the PolyQ diseases and other NMD.

To warrant that advances affect a large fraction of patients we limited the selection to a number of major categories, some of which are in a promising stage of etiological and therapeutic research while some others are in great need of further classification. The efforts will be connected through a NEUROMICS platform for impact, communication and innovation that will provide tools and procedures for ensuring trial-readiness, WP performance, sustainability, interaction with the chosen Support IRDiRC and RD-Connect project and involvement of stakeholders in the NDD/NMD field.

### **Lay Summary**

**Further information available at:**

#### **Types:**

Investments > €500k

#### **Member States:**

European Commission

#### **Diseases:**

Neurodegenerative disease in general

#### **Years:**

2016

#### **Database Categories:**

N/A

#### **Database Tags:**

N/A