NeurOomics: -omics research for diagnosis and therapy in rare neuromuscular and neurodegenerative diseases – an EU funded FP7 project

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Project focus

Neuromics will use the most sophisticated -omics technologies to revolutionise diagnostics and develop pathomechanism-based treatments for ten rare neuromuscular and neurodegenerative diseases affecting primarily the cortex, basal ganglia, cerebellum, spinal cord, peripheral nerves, neuromuscular junction and the muscle.

Diseases covered

<table>
<thead>
<tr>
<th>Disease covered</th>
<th>Disease coordinator in Neuromics</th>
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<tr>
<td>Huntington’s disease</td>
<td>Sarah Tabrizi</td>
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<td>Frontotemporal lobar dementia</td>
<td>Alexis Brice</td>
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<td>Hereditary spastic paraplegia</td>
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<td>Ataxia</td>
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<td>Lower motor neurone disease</td>
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<td>Congenital myasthenic syndrome</td>
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<td>Congenital muscular dystrophies and myopathies</td>
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<td>Muscular dystrophies</td>
<td>Gert-Jan van Ommen</td>
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<tr>
<td>Muscular channelopathies</td>
<td>Michael Hanna</td>
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Approach

Neuromics is a large project with very ambitious objectives. In order to achieve these, the tasks have been divided into 3 scientific ‘sub-projects’ each interlinked and crucial for the others’ success. The first will use existing patient cohorts which are clinically well defined, to identify novel disease causing genes, modifiers and biomarkers. This information will be used by the second ‘sub-project’ which will focus on moving the newly identified genes and biomarkers towards application in clinical practice. The final sub-project focuses on the use of -omics technologies to develop novel therapies using the potential drug-targets identified in other parts of the project. Key to this approach is the partners’ commitment to secure and effective data-sharing.

Neuromics will feed data to the RD-Connect platform and work closely together to shape and develop its suite of tools for rare-disease research.

Neuromics expected impacts

This means that Neuromics research will contribute significantly to the International Rare Diseases Research Consortium’s objectives of a genetic diagnosis for all rare disease patients and the development of 200 new therapies by 2020.

After 18 months

- Whole exome sequencing has been completed for around half of the planned 1100 patient samples
- Analysis of this WES data has so far revealed 33 new disease-causing genes
- Patients have received genetic diagnoses where they did not have one before
- Samples have been collected and identified in biobanks for biomarker research
- Samples have been collected from patients with extremes of phenotype for disease modifier research
- Detailed clinical data has been recorded in a systematic manner as part of deep phenotype and entered into a customised database for use by the consortium
- The Care and Trial Site Registry (CTSR) has been expanded to include neurodegenerative diseases in addition to neuromuscular. The CTSR aims to help the pharmaceutical industry and clinical investigators select trial sites as well as to help to identify potential partners for upcoming research projects
- Procedures have been drafted, agreed and adopted which ensure data-sharing within Neuromics and also with the wider rare-disease community through the RD-Connect platform and via the publically funded European Genome-phenome Archive (EGA)

Next steps

Over the coming months and next stages of Neuromics, the research will move further into analysis of the results from whole exome and RNA sequencing. This will benefit from the ethos of collaborative working and data-sharing established between partners as data from different centres can be mined together.

The work on biomarker discovery and validation will continue and, as new biomarkers correlate to neuromuscular and neurodegenerative disease are confirmed, partners from Neuromics and RD-Connect will work together to ensure their clinical utility and impact on patients.

In a similar way, new gene discoveries will be published and shared with workshops and training courses planned by Neuromics to disseminate new disease information widely within the medical community.

More information

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