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NeuroOmics

Neuromics newsletter

In this issue

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Welcome!

Thanks for your interest in Neuromics and welcome to our first newsletter! You will find here some background about the project and what it hopes to achieve over the next 5 years. We will bring you issues of the newsletter every 6 months with updates on progress, topical articles and relevant news items.

Alongside an introduction to the project, this issue will explain a little about some of the technologies used in this kind of research, particularly –omics technologies. We hope you find it of interest.

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Dear patients, dear colleagues and friends

We hope you have the same spirit and enthusiasm as we do for this project which will definitively guide –omics research in the field for the next 5 years.

With the substantial support of the EU, Neuromics will identify numerous new genes underlying neurological and neuromuscular diseases and we will develop fast and reliable genetic tests to transfer this knowledge into the clinic. We will also develop biomarkers which are very important for future treatment studies and we will undertake pathogenesis research and even first clinical trials.

For the first time, we are able to include several world class leaders from the United States, Canada and Australia as well. Together, we will make a difference!

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What is Neuromics?

Using the most sophisticated -omics technologies, Neuromics will revolutionize diagnosis and develop new treatments for ten major neurodegenerative and neuromuscular diseases affecting the brain and spinal cord, peripheral nerves and muscle.

Rare diseases affect up to 36 million people in Europe

Neurodegenerative and neuromuscular diseases (NDD/NMD) form one of the most frequent groups of rare diseases, affecting the life and mobility of over 500,000 patients in Europe and millions of their caregivers, family members and employers. Neuromics is a research consortium which brings together leading research groups in Europe, highly innovative companies and other experts in the relevant fields.

Conditions included in Neuromics fall into the following categories:

Huntington's disease
Hereditary motor neuropathy
Fronto-temporal lobar dementia
Congenital myasthenic syndrome
Hereditary spastic paraplegia
Congenital muscular dystrophy & myopathy
Ataxia
Muscular dystrophy
Spinal muscular atrophy & lower motor neuron disease
Muscular channelopathy

Specifically, Neuromics hopes to discover more genes which *cause* NDDs and NMDs and other genes which *modify* the diseases and determine how or why they might affect individual patients differently. The discovery of new causative or modifying genes may point to potential novel targets for drug therapy.

The project will also identify and confirm new biomarkers associated with these diseases. Biomarkers are substances in the body that offer a way to measure normal or abnormal processes. This means that they can be useful to accurately measure how a disease is progressing.

Because of this, a major application of new biomarkers will be in clinical trials. If biomarkers can be measured in patients' blood or urine, samples of these can be taken throughout a study. Levels of these biomarkers will clearly and accurately show whether the drug being tested has had an effect or not.

The Neuromics project will take place over 5 years and is funded by the EU until September 2017.

More information about the project background and up to date information can also be found at www.rd-neuromics.eu

Who is involved in Neuromics?

Professor Olaf Riess at the University of Tübingen in Germany coordinates the project

Prof. Brunhilde Wirth, Cologne, and Prof. Gert-Jan van Ommen, Leiden, are co-coordinators

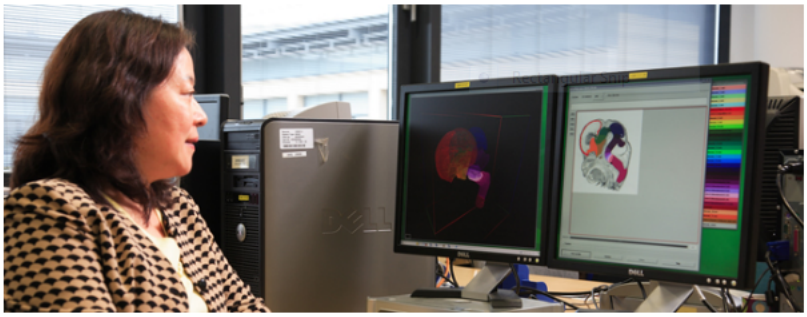
For projects like this to be successful, collaboration of many professionals with different skills is required. There are 19 EU-funded partners involved in Neuromics coordinated by Professor Riess at the University of Tübingen, Germany. The consortium also includes 'associated partners' in the US. These centres will not receive funding from the project but will be working closely with us in order to achieve our common aims.

There is also a Patient Association Committee whose job it is to make sure patients' interests are represented and that they are kept informed about Neuromics' progress. The project's website has full details about the partners and the patients' committee at www.rd-neuromics.eu

NeuroOmics

Integrated European Project on Omics Research of Rare Neuromuscular and Neurodegenerative Diseases

PARTICIPANTS | STRUCTURE | NEWS & EVENTS | LINKED PROJECTS



Concerns raised over draft of EU data protection regulation

Concerns raised over draft of EU data protection regulation: A large group of patient organisations and research consortia involved in the rare disease field has released a public statement expressing deep concern over a draft of the proposed new legal framework for the protection of personal data in the EU, the so-called Data Protection Regulation.
Read more...

Project aims

Neuromics aims to revolutionize diagnostics and develop new treatments for ten major neuromuscular and neurodegenerative diseases.

It will do this by bringing together leading research groups in Europe, five highly innovative SMEs and overseas experts. Together, they will use the most sophisticated -omics technologies in order to:

- increase the number of patients with a genetic diagnosis;
- develop biomarkers for clinical application;
- improve understanding of pathophysiology and identify drug targets;
- identify disease modifiers;
- develop targeted therapies;
- translate findings to other, related disease groups.

Disease fields

Neurodegenerative (NDD) and neuromuscular (NMD) diseases are amongst the most frequent of rare diseases, affecting the life and mobility of more than 500,000 patients and families in Europe.

The focus of Neuromics is on 10 major disease categories. Some of these are at a promising stage of etiological and therapeutic research, whilst in others, diagnostic and therapeutic concepts are still preliminary. The conditions included are:

- Huntington's disease
- Fronto-temporal lobar dementia
- Hereditary spastic paraplegia
- Ataxia
- Spinal muscular atrophy
- Charcot Marie Tooth disease
- Congenital myasthenic syndrome
- Congenital muscular dystrophy and myopathy

Project partners

- **University of Tübingen, Germany - Coordinator**
- Agilent Technologies, Sweden
- Ariadne Diagnostics, LLC, USA
- Bio-Prodict, The Netherlands
- Cambridge University, UK
- Children's National Medical Center, USA
- deCODE genetics, Iceland
- German Center for Neurodegenerative Diseases
- Institut National de la Santé et de la Recherche Médicale, France
- Leiden University Medical Center, The Netherlands
- Newcastle University, UK
- Profilomic, France
- Ste-Justine Hospital Research Center, Canada
- Universitätsklinikum Freiburg, Germany
- Universite D'aix Marseille, France
- University College London, UK
- University Hospital Cologne

The Neuromics website

Neuromics has its own website (www.rd-neuromics.eu) which has full details of the project, progress updates and information about who is involved. There will also be more information for patients on the site very soon which we hope to have translated into languages other than English.

Neuromics is a translational research project – What does this mean?

Scientists are constantly discovering more and more about the human body. We now understand more than ever the processes which keep us alive and well along with some of the things that go wrong causing disease or ill-health. Much of new discovery and technology comes from basic research which takes place in the laboratories of academic institutions or companies around the world. However, harnessing this knowledge and turning new discoveries into useful, marketable treatments of direct benefit to human health is much more difficult.

Translational research bridges the gap between laboratory and patient

Translational research aims to overcome this problem. It is research which turns new knowledge and understanding gained in the laboratory into meaningful therapies, drugs or preventative measures which are of direct benefit to patients. In short, it bridges the gap between laboratory and patient.

One way in which Neuromics does this by trying to identify new genes associated with the cause or progression of rare neuromuscular and neurodegenerative diseases. These new genes will be used to find a diagnosis for more patients and to suggest new areas for potential drug targets. This may lead to new drugs which will be of direct benefit to patients' health.

-omics Research

*A **genome** is all the DNA in an organism. In all living things, it is made from a sequence of just 4 different chemical compounds called **bases**. These bases are referred to as A, T, G and C*

Between 1990 and 2003, the human genome project embarked on the huge task of unlocking the sequence of the entire human genome. This revealed a 3 billion long combination of bases (the simplest bacteria has a sequence of 600,000 in its genome).

Just 2% of this sequence contains our genes – the bits which tell the body how to make the protein building blocks of life. The rest makes up the non-coding regions of the genome. This non-coding part is not fully understood but some of it is responsible for turning the genes on and off and for maintaining the structure of the DNA itself.

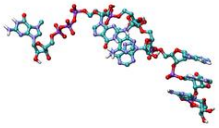
This new knowledge combined with rapid advances in technologies which allow quicker and cheaper sequencing have brought about new ways of doing research. The suffix, -omics is often added to previously existing fields in order to indicate that the research is highly data-rich and on a very large scale. '–omics' involves looking at the whole of something rather than individual parts.

Analysing the genome

Genomics

Analysing RNA

Transcript-omics



Analysing proteins made

Proteomics

For example, genetics becomes genomics when research is no longer looking just at individual genes but instead is studying large amounts of the genetic code – possibly even all of a person's genome.

Genes code for the order of amino acids which join together to build proteins and so the study of these proteins and their role in human genetic diseases is as important as the study of the genes. Proteomics is research which looks at the proteins made and their function as well as at complex interactions between all the proteins in a cell. In the past it may have only been possible to look at individual proteins, one at a time.

Getting from the sequence of bases which makes up a gene to the protein for which it codes requires the copying or transcription of the DNA into RNA. The RNA 'transcripts' of the gene are another area of –omics research – transcriptomics.

Pharmacogenomics is another field of –omics research. It is the study of how a person's genome might affect their response to particular therapies. This is a field which is in its infancy at present, but the hopes are high. Pharmacogenomics may lead to drug treatments tailor-made for individual patients with a reduction in side-effects, an increase in efficacy and therefore, a reduction in costs.

The field of –omics is rapidly increasing with new terms being added to the list all the time. However, all –omic studies have in common their large-scale approach, their view of the whole of something and their handling of very large amounts of data.

Of course, a large amount of data is not necessarily useful in itself. The tools and technologies used to analyse the information generated by –omics research are critical. There have been, and continue to be, huge advances in 'bioinformatics' – the use of software to analyse, organise, store and retrieve biological data.

More information about the human genome project can be found at:
http://www.ornl.gov/sci/techresources/Human_Genome/project/about.shtml

News and Events

The Neuromics team at the project kick-off meeting in Sitges, in January 2013

Neuromics kick-off meeting

The kick-off meeting for the Neuromics project was held in Sitges, just outside Barcelona from 25-27th January. This was a great opportunity for representatives from each of the partners and the patient and ethics boards to get together to discuss the first actions needed for the project and to plan for the longer term.

Neuromics shared this meeting with another '-omics' project which is conducting research into rare kidney diseases – EURenOmics - and the RD-Connect project which will provide an integrated platform for connecting databases, registries, biobanks and clinical bioinformatics for rare disease research. More information about these projects is given below.

RD-Connect has made its presentations from this meeting publically available at: http://rd-connect.eu/meetings/kom-files/kom-presentations.htm

Concern for the future of rare disease research

A large group of patient organisations and research consortia involved in the rare disease field has released a public statement expressing deep concern over a draft of the proposed new legal framework for the protection of personal data in the EU, the so-called Data Protection Regulation.

Please consider writing to your MEP and signing the petition

It is feared that if the regulation is passed by the European Parliament in the wording released on 16 January 2013 by Jan Philipp Albrecht (Greens/European Free Alliance, Germany), this could spell the end of progress in health research in Europe, in particular the end of research into rare diseases.

The ability to share data is considered crucial to advances in medical research into rare diseases, and the group behind today's statement, which includes the voice of patients from three continents, strongly urges the members of the European Parliament who will be considering the proposal to bear in mind and protect the right of patients to quality healthcare and to work on achieving the right balance between data protection and creating a research-friendly environment that will not put future medical advances for rare disease patients at risk.

Please consider:

- Writing to your MEP to express your concerns (find your MEP at http://www.europarl.europa.eu/portal/en)
- Forwarding this information to all those with an interest in rare disease
- Signing the petition at http://www.change.org/en-GB/petitions/don-t-stop-research-on-rare-diseases-2

Neuromics is launched alongside other rare disease projects

Rare diseases – while individually uncommon – affect one person in every 17. Around 80% of rare diseases have a genetic component but many patients do not have a genetic diagnosis. There are few effective therapies.

The EU has now awarded 38 million Euros for research (which includes the Neuromics project) into rare diseases and for the development of a central global rare disease hub (RD-Connect) involving 70 institutions that will allow scientists to share data from their genomics research projects.

This will lead to faster diagnosis and better treatments and will improve the quality of life for patients with rare diseases.

Professor Hanns Lochmüller of Newcastle University, UK, who is leading the new rare disease hub, RD-Connect, said: "Being able to sequence a person's entire genetic code is an important advance, particularly for people living with the many rare genetic disorders, but it has also shown us that sequencing is only the first part of the story. It doesn't replace clinical expertise – in fact, being able to combine genetic data with clinical data is more important than ever."

IRDiRC's goal is to see 200 new therapies and genetic diagnoses for all rare diseases by 2020

The International Rare Diseases Research Consortium (IRDiRC), under which these new grants have been awarded, aims to accelerate research into rare diseases. Professor Paul Lasko of McGill University in Montréal, Canada, Chair-Elect of the IRDiRC Executive Committee, explained: "IRDiRC's goal is to reach 200 new rare disease therapies, and diagnoses for all rare diseases, by the year 2020. To this end, it is launching four major projects which will combine international genetic data with clinical information and data on biomaterials to help interpret the vast amounts of data the genome yields. This will aid scientists in the search for genetic causes of diseases and help identify new ways to create targeted therapies".

The funding awarded supports the following four projects:

- Identifying the genetic and epigenetic causes of rare kidney disorders – EURenOmics led by Heidelberg University Medical Centre, Germany http://www.eurenomics.eu/

- Addressing rare neurodegenerative and neuromuscular disorders using next generation whole-exome sequencing – Neuromics led by the University of Tübingen, Germany http://rd-neuromics.eu/

- Developing a global infrastructure to share the research of rare disease projects – RD-Connect led by Newcastle University, UK http://rd-connect.eu/

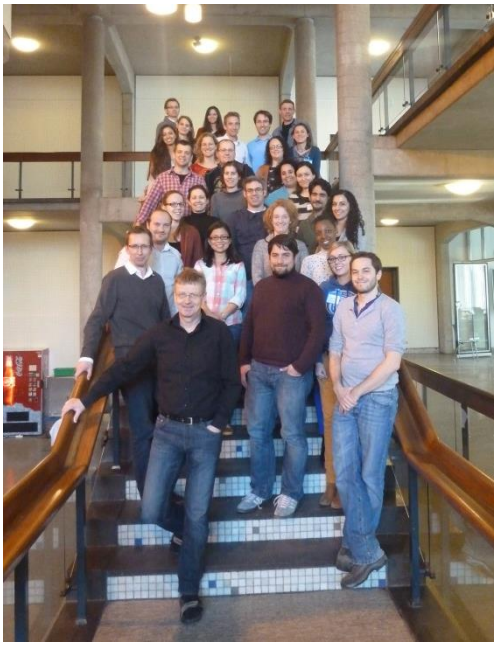
- Supporting international rare disease collaboration through IRDiRC – SUPPORT-IRDiRC led by INSERM, France http://www.irdirc.org/

Neuromics partners attend a sequencing workshop in Cologne

From 27-29 March 2013, 29 participants from Neuromics partner institutions attended a three day workshop on the use of the deCODE Clinical Sequence Miner Software.

During the workshop partners learned how to analyse the data produced by the whole exome sequencing of patients' samples. Within the workshop itself, partners were able to identify the genetic cause in several patients and find promising new clues in others.

The workshop was coordinated by Jóhann Haukur Sigurðsson from deCODE and was hosted by Brunhilde Wirth Cologne.



Diary dates

Visit the Neuromics website for more details of these events: www.rd-neuromics.eu

16th – 17th April 2013
IRDiRC Scientific Conference
Dublin, Ireland

21st – 24th April 2013
MDA Scientific Conference: Therapy Development for Neuromuscular Diseases
Washington, USA

11th – 13th June 2013
SPATAX 2013 Meeting
Paris, France

13th – 16th June 2013
Families of SMA
California, USA

25th – 27th June 2013
5th International CMT Consortium Meeting
Edegem, Belgium

27th – 30th June 2013
PPMD Annual Connect Conference
Baltimore, USA

11th – 12th July 2013
Cure-CMD Family Conference
Bethesda, USA

27th – 29th August 2013
INCT: Neuroinformatics 2013
Stockholm, Sweden

15th – 18th September 2013
2013 World Congress on Huntington's Disease
Rio de Janeiro, Brazil

1st October – 5th October 2013
World Muscle Society 18th International Congress
California, USA

6th – 8th October 2013
9th Annual Meeting of the Oligonucleotides Therapy Society
Naples, Italy

5th October 2013
Muscular Dystrophy Campaign Annual Scottish Conference
Glasgow, UK

12th October 2013
Muscular Dystrophy Campaign Annual Conference
Nottingham, UK

Please send dates for the diary to catherine.turner@ncl.ac.uk