## 1. Report on Research Projects and Clinical Trials of last five years

### 1. A) Relevant clinical trials of last 5 years

Universitätsklinikum Tübingen

| Title | Objective | Multi- or monosite | Sponsor |
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| - A phase I/II, multicenter, open-label, controlled dose escalation study of HGT-1110 administered intrathecally in children with metachromatic Leukodystrophy | To determine the safety of ascending doses of HGT-1110 administered by intrathecal (IT) injection for 40 weeks in children with metachromatic leukodystrophy (MLD).<br>• To evaluate the clinical activity of IT administration of HGT-1110 on gross motor function<br>• To assess single and repeated-dose pharmacokinetics of HGT-1110 in serum<br>• To assess concentrations of HGT-1110 in cerebrospinal fluid (CSF) | Multisite | Shire 2013-2016 |
| Dysport RU | Evaluation of Efficacy and Safety of Dysport and Dysport RU to Placebo of Subjects with CD | Multisite | IPSEN GROUP |
| TOWER | Evaluation of Efficacy and Safety of Xeomin for treatment of spasticity | Multisite | MERZ |
| CD-NIS-longterm | Evaluation of the treatment effect of Dysport in CD after two cycles in naïve and pre-treated patients | Multisite | IPSEN GROUP |
| Interest in CD 2 | Evaluation of longterm response to Botulinumtoxin type A in subjects suffering from idiopathic CD-Pharmaco-economic impact | Multisite | IPSEN GROUP |
| Botox Prophylaxis in Chronic Migraine | Observational Study in selected European Countries | Multisite | ALLERGAN |
| AS-NIS early BIRD | Investigation of Effectiveness of Botu-linumtoxin A (Dysport) Injections In Patients suffering from Post-stroke arm s with respect to early, medium or late start o treatment | Multisite | IPSEN GROUP |
| BoTN for quality of life in different dystonia syndromes | Quality of life in different dystonia syndromes and post-stroke spasticity before and after botulinumtoxin treatment | Multisite | MERZ |
| Multisyn | Multimodal imaging of rare Synucleinopathies | Multi | EU & Affiris |
| Mito-PD | Evaluation der mitochondrialen endo-Phänotypen der Parkinson-Krankheit | Multi | BMBF |
| Promesa | Study to investigate the effect of EGCG supplemantation on disease | Multi | LMU München |

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| | progression of patients with Multiple System Atrophy (MSA) | | |
| MIGAP | Markers in GBA-associated PD. Verlaufsuntersuchung des DZNE zur Detektion von Biomarkern, welche Unterschiede zwischen Parkinson Patienten mit und ohne GBA-Mutation, sowie asymptomatischen Gesunden mit und ohne GBA-Mutation aufzeigen soll. | Multi | DZNE |
| RIVA | Exelon at dementing syndrome of patients with supranueclear palsy | Mono | Novartis GmbH & Uniklinik Tübingen |
| PSP-Noscira | Study to evaluate the safety, tolerability and efficacy of two different oral doses of NP031112, a GSKInhibitor, versus placebo in the Treatment of patients with mild to moderate progressive supranuclear palsy | Multi | Noscira |
| MSA-RAS 202 | Clinical Trial to Assess the Efficacy, Safety, and Tolerability of Rasagiline Mesylate 1 mg in Patients with Multiple System Atrophy of the Parkinsonian Subtype (MSA-P) | Multi | TEVA |
| EPI589-15-002 | Safety and Biomarker Study of EPI-589 in Mitochondrial Subtype and Idiopathic Parkinson's Disease Subjects | Multi | Edison |

Medizinische Universität Innsbruck

| Title | Objective | Multi- or monosite | Sponsor |
|---|---|---|---|
| Safety and Tolerability of Carbamylated Erythropoietin in Friedreich's Ataxia | Safety and tolerability of CEPO in FRDA (Phase II) | Multi-center, CRT | Lundbeck |
| Randomisierte, placebokontrollierte Doppelblindstudie der Phase III zur Wirksamkeit, Sicherheit und Verträglichkeit von Idebenone bei der Behandlung von Patienten mit Friedreich-Ataxie. (MICONOS) | Wirksamkeit, Sicherheit und Verträglichkeit von Idebenone bei der Behandlung von Patienten mit Friedreich-Ataxie (Phase III). | Multi-center | Santhera |
| Eine offene Verlängerungsstudie der Phase-III zur Erhebung von Langzeitdaten zur Sicherheit und Verträglichkeit von Idebenone bei der Behandlung von Patienten mit Friedreich-Ataxie (FRDA) | Langzeitdaten zur Sicherheit und Verträglichkeit von Idebenone bei der Behandlung von Patienten mit Friedreich-Ataxie (FRDA) | Multi-center | Santhera |

Charles University Prague

| Title | Objective | Multi- or monosite | Sponsor |
|---|---|---|---|
| A Multicenter, Multinational, Randomized, Double-Blind, Placebo-Controlled,Parallel-Group Study to Evaluate the Efficacy and Safety of Laquinimod (0.5,1.0 and 1.5 mg/day) as Treatment in Patients with Huntington's Disease | Phase II (2015-) | multisite | Teva |
| An intranational observational Study on long term response to BTX injections in subjects suffering from idiopathic cervical dystonia | Phase III (2014 –) | Intranational Multisite | IPSEN PHARMA |

University Hospital Bordeaux

| Title | Objective | Multi- or monosite | Sponsor |
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| BIOAMS | Fluid biomarker development | Monosite | University Hospital Bordeaux |
| BIOPARK | Fluid biomarker development | Multisite | French Health Ministry |
| COGAMS | Assessment of cognition | Monosite | University Hospital Bordeaux |
| DOPS-AMS | Treatment trial | Multisite | French Health Ministry |
| AZD3241 | Treatment trial | Multisite | Astra Zeneca |
| SYMPATH | Treatment trial | Multisite | FP-7 |
| MULTIPAMS | Imaging biomarker development | Monosite | INSERM (Institut National de la Santé et de la Recherche Médicale) – DGOS (French Health Ministry) |
| MSA-FLUOXETINE | Treatment trial | Multisite | French Health Ministry |
| MSA-DOUL | Pathophysiology of pain in MSA | Monosite | Fondation de France |
| UROPARKTENS | Treatment trial | Multisite | French Health Ministry |
| SEROTAMS | Imaging of serotonin system | Multisite | French Health Ministry |
| HYPOSOMPARK | Pathophysiology of postprandial OH | Multisite | French Health Ministry |
| MSA rasagiline | Treatment trial | Multisite | TEVA Pharma |

Reference Centre for Rare Diseases 'Neurogenetics', Pitié-Salpêtrière Hospital

| Title | Objective | Multi- or monosite | Sponsor |
|---|---|---|---|
| Evolution of albumin on AOA1 patients supplemented with Coenzyme Q10_ AOA1 | To compare the evolution of 12-months CoQ10 supplementation with that of placebo on AOA1 patients' albumin level | Multi -site | AP-HP |
| Therapeutic Metabolic | To study the efficacy of three candidate | Monosite | COSSEC, |

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| Intervention in Patients With Spastic Paraplegia SPG5_SPA-M | molecules in order to decrease the production of oxysterols by reducing the synthesis of cholesterol and/or regulate the production of bile acids and/or enabling neuroprotective action within the motor neuron. | | Carnot, ASL |
| Phase II Open Label Study Using Triheptanoin in Patients With Glucose Type 1 Transporter Deficiency_GLUTHEP | To study the efficacy of triheptanoin oil in patients with GLUT1 deficiency syndrome. | Monosite | Ultragenyx |
| Triheptanoin Treatment Trial for Patients With Adult Polyglucosan Body Disease_GLUCOHEP | To determine if triheptanoin is an effective treatment for the symptoms of Adult Polyglucosan Body Disease. | Multisite | Ultragenyx |
| SPIDY 3 Pallidal and additional target (thalamus or sub-thalamic nucleus) in dystonia | To determine the effect of additional target in<br>Complement with "classical " pallidal DBS<br>(deep brain stimulation) in dystonia | Multisite | PHRC |
| A randomized, controlled, double-blind, crossover trial of zonisamide in myoclonus-dystonia. (ESPSILON-ZETA) | To determine the beneficial effect of zonizamide<br>On myoclonus and dystonia in myoclonus-dystonia<br>Double blind randomized placebo controlled,<br>Cross over pharmacological study | Monosite | PHRC |

National Reference Center For Huntington's Disease

| Title | Objective | Multi- or monosite | Sponsor |
|---|---|---|---|
| Multicentre trial of treatment of Huntington's disease by cysteamine ( CYST HD) | neuroprotection | Multisite | DRC ANGERS |
| Extension of the multicentric intracerebral grafting trial in HD | Long lasting fllow-up of transplanted patients using fetal cells | Multisite | APHP |
| NEURO HD | Comparison of neuroleptics | Multisite | APHP |
| PRIDE HD | Pridopidine efficacy | Multisite | TEVA |
| A multi-centre, multi-national prospective observational imaging biomarker study in early stage Huntington's disease (HD) patients to assess imaging techniques and parameters able to support efficacy studies with SEN0014196 in HD patients during Phase II and III studies | neuroprotection | Multisite | FP7 |

Universitätsklinikum Bonn

| Title | Objective | Multi- or monosite | Sponsor |
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| A phase III double-blind, randomised, placebo-controlled study of the efficacy, safety and | see title | multicentre | Santhera |

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| tolerability of idebenone in the treatment of FRDA (MICONOS) | | | |
| Randomised, double-blind, placebo-controlled study of Lu AA24493 (CEPO) in patients with FRDA | see title | multicentre | Lundbeck |
| Effects of acetyl-DL-leucine on cerebellar ataxia - a multinational, multicenter, randomized, double-blind, placebo-controlled, 2-way crossover phase III trial (ALCAT) | see title | multicentre | University of Munich |

Institute of Neurogenetics, University of Lübeck

| Title | Objective | Multi- or monosite | Sponsor |
|---|---|---|---|
| Medivation Dimebon Extension Studie | treatment of Huntington disease | multi | Medivation |
| Double blind, randomised, prospective placebo controlled parallel group phase III study to investigate the effect of EGCG supplementation on disease progression of patients with multiple system atrophy (PROMESA) | treatment of MSA | multi | Investigator initiated |
| A phase 2, randomized, placebo controlled double blind proof-of-concept study of the efficacy and safety of PF-02545920 in subjects with Huntington's disease | treatment of Huntington disease | multi | Pfizer |
| A dose escalating, proof-of-concept, phase IIa study to investigate the safety and tolerability, the pharmacokinetic and the pharmacodynamic of BN82451B, administerd twice daily over 4 weeks, in male patients with Huntington's disease | treatment of Huntington disease | multi | Ipsen |
| Mito-PD: An omics-based strategy using coenzyme Q10 in patients with Parkinson's disease: Concept evaluation in a double-blind randomized placebo-controlled parallel group trial | Treatment of PD | multi | BMBF |

Klinikum der Ludwig- Maximilians-Universität München

| Title | Objective | Multi-/monosite | Sponsor |
|---|---|---|---|
| A Two-arm Efficacy and Safety Study of Deferiprone in Patients With Pantothenate Kinase-associated Neurodegeneration (PKAN) | To test the efficacy of deferiprone to lower brain iron and to improve the clinical phenotype in PKAN (results in early 2017) | multicentric (as Principal Investigator) | EU-FP7 |
| Long-term Deferiprone Treatment in Patients With Pantothenate Kinase-Associated Neurodegeneration (TIRCON-EXT) | Extension study of the above | multicentric (as Principal Investigator) | ApoPharma |
| ALCAT | (results in 2017) | multicentric (as Investigator) | BMBF |
| Safety and tolerability of carbamylated erythropoietin in Friedreich's ataxia | published (Boesch et al, 2014) | multicentric (as Investigator) | H. Lundbeck A/S |
| A randomized trial of 4-aminopyridine in EA2 and related familial episodic ataxias | published (Strupp et al, 2011) | monocentric (as Investigator) | |

University Hospitals Leuven

| Title | Objective | Multi- or monosite | Sponsor |
|---|---|---|---|
| A Randomized, Double-Blind, Placebo-Controlled, Dose-Escalating, Phase 2a Safety, Tolerability, and Pharmacodynamic Study of Two Doses of an Histone Deacetylase Inhibitor (FRM-0334) in Subjects with Prodromal to Moderate Frontotemporal Dementia with Granulin Mutation Eudract 2014-001489-85 | Evaluate the safety and tolerability of 2 fixed doses of FRM-0334 (300 and 500 mg daily in 2 sequential periods) over 28 days in subjects with prodromal to moderate FTD-GRN<br>• Assess the PD effects of FRM-0334 on the change from baseline in plasma concentrations of PGRN after 28 days | Multisite | Forum Pharmaceuticals |
| 18F-THK5351 tau PET imaging in Alzheimer's disease, frontotemporal degeneration and healthy older adults EudraCT Number: 2014-002976-10 | To investigate the novel tau PET ligand $^{18}$F-THK5351. To determine the brain imaging metrics of the novel tau PET ligand in clinically probable AD and age-matched controls and its capacity to differentiate between AD and healthy controls and between different subtypes of primary progressive aphasia. To interpret the in vivo results in the light of ex vivo binding characteristics of THK5351 for tau aggregates in AD and FTLD subtypes postmortem | Monocentric | KU Leuven |
| Randomized comparison of integrated versus consecutive | To evaluate the effectiveness and safety of integrated | Multisite | Jacques and Gloria Gossweiler |

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| dual task practice in Parkinson's disease: the DUALITY trial.<br>Clinicaltrials.gov NCT01375413. | versus consecutive task training in patients with Parkinson's disease | | Foundation |

Semmelweis University Budapest

| Title | Objective | Multi- or monosite | Sponsor |
|---|---|---|---|
| A Multinational Multicentre Randomized Double-Blind Placebo- Controlled Parallel Group Study to Assess the Efficacy Tolerability and Safety of Talampanel in Subjects with Amyotrophic Lateral Sclerosis ALS. | ALS | Multicentre | TEVA |
| Efficacy and safety of a normal human immunglobulin product for intravenous administration in the treatment of polymyositis and dermatomyositis prospective, randomised, double blind, placebo controlled study | Polymyositis, dermatomyositis | Multicentre | Orfagen |
| Comparison of QUTENZA and pregabalin in painful peripheral neuropathy (ELEVATE) | Painful peripheral neuropathy | Multicentre | Astellas Pharma |
| Multicenter, double-blind placebo controlled trial to analyze the effectivity and tolerability of BAF312 (CL2-78989-010 and CBAF312X2206) | Polymyositis | Multicentre | Novartis Pharma |
| Randomized double-blind proof-of-concept effectivity study with subcutaneous gevokizumab in polymyositis or dermatomyosistis | Polymyosistis/Dearmatomyositis | Multicentre | Servier |

Istituto Neurologico Carlo Besta, Milan

| Title | Objective | Multi- or monosite | Sponsor |
|---|---|---|---|
| Randomized, Double-Blind, Placebo Controlled, Parallel-Group Study to Evaluate the Safety and Clinical Effect of Laquinimod (0.5, 1.0 and 1.5 mg/day) as Treatment in Patients with Huntington's Disease | Evaluate the Safety and Clinical Effect | Multicenter, Multinational | TEVA |
| A Phase IIa Study in Huntington's Disease Patients to Determine the Safety and Tolerability of SEN0014196 (EudraCT 2011- | Determine the Safety and Tolerability | Multicenter, Multinational | SIENA-BIOTECH |

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| 001131-23) | | | |
| Randomised, double blind, placebo controlled study of LuAA24493 in patients with Friedreich's Ataxia to evaluate safety and tolerability and to explore efficacy. (EudraCT No. 2008-003662-2) | Evaluate safety and tolerability and to explore efficacy | Multicenter, Multinational | LUNDBECK |
| A Double-Blind, Placebo-Controlled, Randomized, Parallel Group, 12-Month Safety and Efficacy Trial of Leuco-methylthioninium bis (hydromethanesulfonate) in Subjects with Behavioral Variant Frontotemporal Dementia (bvFTD)<br>STUDY CODE: TRx-237-007 | Evaluate safety and tolerability and to explore efficacy | Multicenter, International | TauRx Therapeutics LTD, Singapore<br>(2014-2015) |
| A Multi-Center, Open-Label Study Evaluating the Safety, Tolerability, and Efficacy of Pridopidine in Patients with Huntington's Disease (Open PRIDE-HD) | Evaluate the Clinical Effect and safety | Multicenter, Multinational | TEVA |
| A randomized, double-blind, placebo-controlled trial of deferiprone<br>in patients with pantothenate kinase-associated neurodegeneration<br>(PKAN) | To explore efficacy | Multicenter, International (4 countries) | ApoPharma<br>EudraCT number: 2012-000845-11 |
| Long-term safety and efficacy study of deferiprone in patients with<br>pantothenate kinase-associated neurodegeneration (PKAN) 4 countries | Evaluate safety and tolerability | Multicenter, International (4 countries | ApoPharma<br>EudraCT<br>No.2014-001427-79 |

Bambino Gesù Children's Research, Rome

| Title | Objective | Multi- or monosite | Sponsor |
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| Iron-related MRI<br>images in patients with pantothenate kinase-associated neurodegeneration (PKAN) treated with deferiprone: results of a phase II pilot trial | Reduction of iron detected<br>by MRI in patients treated by deferiprone | Multi (2 sites) | No profit |

University Medical Center Groningen

| Title | Objective | Multi- or monosite | Sponsor |
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| Ketogenic diet in GOSR2 mutations | To study effectiveness ketoenic<br>diet in progressive myoclonus ataxia | mono | Internal grant |
| Citalopram trial in dystonia | To study the effect of SSRI on dystonia | multi | Internal grant University |
| Physiotherapy trial in dystonia | To study the effect of<br>physiotherapy in dystonia | multi | Jacques and Gloria Gossweiler Foundation & Fonds Nuts-Ohra & Stichting wetenschapsfonds dystonie |

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| Treating Iron Related Childhood Onset Neuro-degeneration | Treating Iron Related Childhood Onset Neurodegeneration | Multi TIRCON | EU KP7 Health |
| Registry / Enroll | To study prospectively the clinical progression of Huntington's disease disease | Multi | European Huntington's Disease Network (EHDN) |

Radboud University Medical Center, Nijmegen, The Netherlands

| Title | Objective | Multi- or monosite | Sponsor |
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| ALCAT | Effect of leucine in ataxia | Multisite | Univ. München |
| Dystonia | Effect of physiotherapy in cervical dystonia | Multisite | School of Amsterdam, Dutch Dystonia Society, Prinses Beatrix Fonds |
| Functional effects of botulinum toxin in the calf muscles of hereditary spastic paraplegia (Feboch-I) | Understanding the effects of botulinum toxin injections on calf muscle strength, spasticity, dynamic balance and gait, with emphasis on the sagittal plane. | Monosite | Ipsen |
| Functional effects of botulinum toxin in the hip adductors of hereditary spastic paraplegia (Feboch-II) | Understanding the effects of botulinum toxin injections on hip adductor strength, spasticity, dynamic balance and gait, with emphasis on the frontal plane. | Monosite | Merz |
| Sjogren-Larsson Syndrome: effects of Zileuton | Effects of Zileuton in SLS | Monosite | Radboudumc / Metakids |

Pitié-Salpêtrière hospital, Paris, France, Reference center for rare dementias

| Title | Objective | Multi- or monosite | Sponsor |
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| PIVOINE FRM-0334-002 | Treatment of FTD patients carrying PGRN mutations | International, multisite | Industrial (FORUM) |
| STIM-LANG | Evaluation of TMS in patients with FTD, PSP and PPA | Monosite | *eye-brain* |

Vall Hebron University Hospital

| Title | Objective | Multi- or monosite | Sponsor |
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| Multicentric, open, expanded access study for the use of RAD001, in patients with subependymal giant-cell astrocitoma (SEGA) associated with tuberous sclerosis complex (TSC). (CRAD001-mic02) | To evaluate safety of RAD001 in patients with TSC and associated SEGA | Multisite | Novartis Pharma |
| A disease international registry to collect data on manifestations, interventions and | To learn about natural history and | Multisite | Novartis Pharma |

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| outcome in patients with TSC –TOSCA study (CRAD001-mic03) | Intervention outcomed in a TSC cohort | | |
| Intravenous immunoglobulin for preparing myasthenia gravis patients for thymectomy and other surgical procedures preventing myasthenic crisis. (MG2012PREP) EudraCT # 2012-001544-21. PI: J.Gámez | IGIV for preparing myasthenia gravis patients for thymectomy and other surgical procedures preventing myasthenic crisis. | Multisite | CSL Behring LLC |
| Multicentric, double blind, randomised, placebo-controlled study of the efficacy and safety of Dexpramipexol in ALS subjects. EMPOWER study. | To evaluate the efficacy, compared to placebo, of a 12-month oral administration of despramipexol 150mg (twice a day) in patients with ALS | Multisite | Biogen Idec Inc. |
| Estudio de extensión, multicéntrico y abierto, para evaluar la seguridad y la eficacia a largo plazo de dexpramipexol (BIIB050) en sujetos con esclerosis lateral amiotrófica. Estudio ENVISION. | Evaluar el perfil de seguridad a largo plazo del dexpramipexol en 850 pacientes con ELA que han finalizado el estudio CL211 o el estudio 223AS302. | Multisite | Biogen Idec Research Ltd |
| Estudio prospectivo, Fase II, multicéntrico, randomizado, doble ciego, controlado con placebo, de grupos paralelos para comparar la eficacia y seguridad de masitinib versus placebo en el tratamiento de pacientes con esclerosis lateral amiotrófica (ELA). | Evaluar la eficacia de masitinib en pacientes de 18-80 años con ELA en tratamiento con riluzol. | Multisite | AB SCIENCE |
| Estudio de fase II/III, aleatorizado, doble ciego y controlado con placebo para evaluar la eficacia y la seguridad de ISIS 420915 en pacientes con polineuropatía amiloide familiar (Familial Amyloid Polyneuropathy, FAP) | Evaluar la eficacia de ISIS 420915 en pacientes con FAP en fase 1-2 que pueden haber recibido tratamiento con tafamidis | Multisite | Ionis Pharmaceuticals, Inc |
| Estudio de extensión abierto y multicéntrico para evaluar la seguridad y eficacia a largo plazo de IgPro20 como tratamiento de mantenimiento de la polineuropatía desmielinizante inflamatoria crónica (CIDP), en pacientes que hayan completado el estudio IgPro20_3003. | Evaluar la seguridad de IgPro20 en pacientes con PDIC que han participado en el ensayo IgPro20_30 03 (el 44/2012 en nuestro centro). | Multisite | CSL Behring GmbH |
| Estudio de SOM0226 en pacientes con polineuropatía amiloide familiar (FAP) y portadores asintomáticos para evaluar la estabilización de la proteina TTR. | Evaluar la eficacia de SOM0226 en pacientes con FAP y V30M<br><br>TTR mutado asintomáticos o en estadio 1-2 y en voluntarios sanos con TTR | Monosite | SOM Innovation Biotech SL (SOM Biotech) |

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| | no mutado. | | |
| Ensayo de extensión, abierto, de fase III de ECU-MG-301 para evaluar la seguridad y la eficacia de Eculizumab en sujetos con miastenia gravis generalizada (MGg) resistente al tratamiento. | Evaluar la seguridad de ecu lizumab en pacientes con miastenia gravis resistente al tratamiento que participaron en el ensayo previo que se trataba de un ensayo clínico enmascarado a doble ciego y controlado con placebo | Multisite | Alexion Pharma EMEA GmbH |
| Estudio de extensión abierto para evaluar la seguridad y eficacia a largo plazo de ISIS 420915 en pacientes con polineuropatía amiloide familiar (PAF). | To evaluate….. | Multisite | Isis Pharmaceuticals, Inc. |
| A double-blind, randomised, placebo controlled study, with three treatment arms, on the efficacy and safety of two everolimus trough level ranges as a coadjuvant treatment in TSC patients suffering partial-onset refractory seizures (CRAD001m2304). | To evaluate everolimus efficacy as coadjuvant in patients with TSC with partial-onset refractory seizures and receiving AEDs | Multisite | Novartis Pharma |
| Nº EudrCT: 2010-018283-16<br>A Randomized, Double-Blind, Placebo-Controlled Phase 3 Study to Investigate the Efficacy and Safety of Progesterone in Patients with Severe Traumatic Brain Injury | Study to Investigate the Efficacy and Safety of Progesterone in Patients with Severe Traumatic Brain Injury | Multisite | Pharmaceutical Research Associates España SAU |
| Nº EudrCT: 2009-012338-56<br>An exploratory phase Iia study to assess safety, tolerability, pharmacodynamics and pharmacokinetics of VAS203 in patients with moderate and severe traumatic brain | Study to assess safety, tolerability, pharmacodynamics and pharmacokinetics of VAS203 in patients with moderate and severe traumatic brain injury | Multisite | Vasopharm GmbH |
| Nº EudrCT: 2007-005802-38<br>Efficacy and Saffety of AP12009 in adults patients with Recurrent or refractory Anaplastic Astrocytoma (WHO grade III) as compared to standard treatment with Temozolamide or BCNU: A Randomized , Activelycontroled, open label clinical phase III study | Efficacy and Saffety of AP12009 in adults patients with Recurrent or refractory Anaplastic Astrocytoma (WHO grade III) as compared to standard treatment with Temozolamide or BCNU | Multisite | Antisense Pharma GmbH |

**1. B) Relevant research projects of last 5 years** (title, funding source)

Universitätsklinikum Tübingen

| Title | Funding source |
|---|---|
| - LeukoTreat WP1 Characterizing leukodystrophies for therapies | EU funds Call identifier FP7-HEALTH-2009-single-stage |
| - Increasing diagnostic accuracy for Niemann Pick C by combining biochemical, genetic and imaging biomarkers | Actelion 2013-2016 |
| COST Action: Europan Network for the Study of Dystonia | EC |
| Whole exome sequencing in patients with myoclonus dystonia | DMRF |
| Genetic basis of dystonia in Turkey | DFG |
| Genetic research in Turkish families with dystonia | MWK |
| Mefopa: Mendelian Forms of Parkinson's Disease | EU |
| MultiSyn: Multimodal Imaging of rare Synucleinopathies | EU |
| MitoPD: Mitochondrial endophenotypes of Parkinson's Disease | BMBF |
| MIGAP: Markers in GBA-associated PD (MIGAP) - early detection, progression, mechanisms, protection | DZNE |
| STN+SNr multicenter Trial: „Combined stimulation of the subthalamic nucleus and the substantia nigra pars reticulata for the treatment of freezing of gait. A phase IIb trial. Tübingen University; Active (recruiting). | Investigator-initiated (third-party funding by Medtronic) |
| Early Stim: European Multicenter study on "The Effect of Deep Brain Stimulation of the Subthalamic Nucleus (STN-DBS) on Quality of Life in Comparison to Best Medical Treatment in Patients with Complicated Parkinson's Disease and Preserved Psychosocial Competence"; Completed. Published in the New England Journal of Medicine. | Investigator-initiated (third-party funding by Medtronic) |
| Early Stim Post Study Follow-up | Investigator-initiated (third-party funding by Medtronic) |
| Duodopa Study 187.3.001: A randomized, double blind, double-dummy, efficacy, safety and tolerability study of levodopa-carbidopa intestinal gel in levodopa responsive Parkinson's disease subjects. Receiving optimized treatments with Parkinson medicinal products who continue to experience persistent motor fluctuations. Published in Lancet Neurology. | Abbvie |
| 03/2014 Combined interleaved stimulation of STN and SNr for mobility impairment related to freezing of gait: design of a randomized controlled phase IIb clinical trial, 03/2014 | EKUT is coordinating center, investigator initiated trial (third-party funding by Medtronic) |

Charles University Prague

| Title | Funding source |
|---|---|
| Biomarkers of progression and treatment response in neurodegenerative disorders | Ministry of Health of the Czech republic (grant nr. 15-25602A) (2015-2020) |
| REM sleep behavior disorder: predicting the of neurodegeneration | GAČR, 16-07879S, (2016-2018) |

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| Effect of specific rehabilitation on gait stability in Huntingtons disease | GAUK 1888214, (2014-2016) |
| Speech disorders and analysis of their mechanisms in Parkinson's disease and other movement disorders. | AZV, 15-28038A, (2015-2019) |
| Analysis of movement disorders for the study of extrapyramidal diseases mechanisms using motion capture camera systems | AZV, 16-28119A, (2016-2019) |
| Diagnostic neurophysiological and laboratory markers and pathophysiological mechanisms of functional movement disorders | AZV, 16-29651A, (2016-2019) |
| REM sleep behavior disorder – clinical presentation, disease course and its implications | AZV, 16-28914A, , (2016-2019) |
| Neuropsychiatric aspects of neurodegenerative diseases | MŠMT, VZ MSM0021620849 (2007-2013) |
| Diagnostic markers and pathophysiological mechanisms of atypical parkinsonian syndromes | IGA MZ ČR, NT 12288-5/2011, (2011-2015) |
| Pathophysiology of neuropsychiatric disorders and clinical applications | MŠMT, VZ 6206094/21620816, (2005-2001) |
| European Network of Reference for Rare Pediatric Neurological Diseases | EU PHEA, nEUroped, (2008-2011) |
| Gait and balance disorders in Parkinson´s disease and atypical parkinsonism - analysis of mechanism and risk factors of falls and development of preventive neurorehabilitation programme | IGA MZ ČR, NS10336-3/2009, (2009-2011) |
| Pathophysiological mechanisms of neuromodulation therapy in dystonia | IGA MZ ČR, NT 12282-5/2011, /2011-2015) |
| Multidisciplinary approach in the diagnosis of frontotemporal lobar degenerations and tauopathies: new insights into pathogenetic mechanisms | GA MZ ČR, NT12094-5/2011, (2011-2014) |
| European Network and Registry for Homocystinurias and Methylation Defects | EAHC |
| The International Niemann-Pick Disease Registry | EAHC |
| Biochemistry and Cell Biology of Human Acetyl-Coenzyme A: Alpha-Glucosaminide N-Acetyltransferase – Basis for Future Therapeutic Applications in Mucopolysaccharidosis type IIIC | NS10342 |
| Identification of the genetic and molecular basis of rare genetic disorders using novel genomic methods | NT13116 |
| Niemann-Pick disease type C : clinical, molecular genetic, biochemical and morphological study. Development of new diagnostic and predictive algorithms | NT12239 |
| Characterization of the first mouse model of mucopolysaccharidosis type IIIC | NT13122 |
| Phenotype-genotype relations in lysosomal storage diseases related to beta-galactosidase deficiency | MEB060904 |
| Progress in methods of laboratory diagnostics of inherited lysosomal neurodegenerative disorders | NT14015 |
| Regulation of the gene expression and new approaches to the molecular diagnostics of the ornithine transcarbamylase deficiency | NR9364 |
| New screening system of genetic defects of the de novo purines synthesis and its application in differential diagnosis of patients with psychomotor retardation of unknown etiology | LH11031 |
| Hydrogen sulfide metabolism in homocystinurias | LD14082 |
| Optimalization of Laboratory Newborn Screening of Inherited Metabolic Disorders | NT12213 |

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| Plasma activities of intracellular enzymes in selected inborn errors of metabolism - suitability for diagnosis and for predicting responsiveness to therapy | NT14159 |
| European Network and Registry for Homocystinurias and Methylation Defects | EAHC |
| The International Niemann-Pick Disease Registry | EAHC |
| Biochemistry and Cell Biology of Human Acetyl-Coenzyme A: Alpha-Glucosaminide N-Acetyltransferase – Basis for Future Therapeutic Applications in Mucopolysaccharidosis type IIIC | NS10342 |
| Identification of the genetic and molecular basis of rare genetic disorders using novel genomic methods | NT13116 |
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Centre of Hereditary Ataxias, Motol University Hospital, Prague, Czech Republic

| Title | Funding source |
|---|---|
| Grant TA03011175 (2013-2016, TA0/TA) - Development of rehabilitation and neurology diagnostic tools for using 3D motion analysis | TA0 - Technology Agency of the Czech Republic (TAČR) |
| Grant MZCR NT 13120-4/2012 (2012-2015). Disease caused by mutations in the MECP2 gene in the Czech Republic and the influence of other genetic determinants of their phenotypic manifestations. | Ministry of Healthy CR |
| Developmental Program of fields of science at Charles University (PRVOUK) - P38; 2012-2015. Biological aspects of human exploration of movement. | Charles University |
| IGA MZ ČR: NS/100005-4 2008 – 2011 Hereditary spinocerebellar ataxia - extending molecular genetic analysis of continuous longitudinal multidisciplinary studies of patients' families | Ministry of Healthy CR |
| Part of the research project University Hospital Motol (2012- 2017): Neurogenetic problems spinocerebellar and neurodegenerative diseases | Ministry of Healthy CR |

French reference Center for MSA (Toulouse and Bordeaux site)

| Title | Funding source |
|---|---|
| BIOAMS | University Hospital Bordeaux |
| BIOPARK | French Health Ministry |
| COGAMS | University Hospital Bordeaux |
| DOPS-AMS | French Health Ministry |
| AZD3241 | Astra Zeneca |
| SYMPATH | FP-7 |
| MULTIPAMS | INSERM (Institut National de la Santé et de la Recherche Médicale) – DGOS (French Health Ministry) |
| MSA-FLOUXETINE | French Health Ministry |
| MSA-DOUL | Fondation de France |
| UROPARKTENS | French Health Ministry |
| SEROTAMS | French Health Ministry |
| HYPOSOMPARK | French Health Ministry |
| MSA rasagiline | TEVA Pharma |

Reference Centre for Rare Diseases 'Neurogenetics', Pitié-Salpêtrière Hospital

| Title | Funding source |
|---|---|
| Prospective study of individuals at risk for spinocerebellar ataxia type 1, type 2, type 3 and type 6 (SCA1, SCA2, SCA3, SCA6)_ RISCA | Erare |
| European Friedreich's Ataxia Consortium for Translational Studies_EFACTS | FP7 |
| Biomarkers in Autosomal Dominant Cerebellar Ataxia_ BIOSCA | PHRC |
| Integrated European Project on Omics Research of Rare Neuromuscular and Neurodegenerative Diseases_NEUROMICS | FP7 |
| Polyglutamin diseases, cancers and cardiovascular diseases_MEPKA | Institut Curie |
| Identification of genetic, epigenetic and environmental modifiers in hereditary spastic paraplegia_MODIFSPA | VERUM |
| Longitudinal analysis of oral communication in Friedreich's ataxia_ ORFA | AFAF |
| SPIDY 3 Pallidal and additional target (thalamus or sub-thalamic nucleus) in dystonia | PHRC |
| A randomized, controlled, double-blind, crossover trial of zonisamide in myoclonus-dystonia. | PHRC |
| Consolidation of motor memory in writer's cramp patients | AMADYS |
| CERDYS : cerebellar control of cortical plasticity in writer's cramp | AMADYS |
| Gamma: implication of the cerebellum in sensori-motor adaptation in dystonia: gamma activity | AREVA |
| CERTRE Intrinsic signature of essential tremor in the cerebello-frontal network | APTES |
| Experimental therapeutics and pathophysiology in orthostatic tremor: effect of cerebellar repetitive TMS in primary orthostatic tremor: clinical, electrophysiological and MRI study | APTES |
| European project and mendolian forms of Parkinson disease_MEFOPA | FP7 |

National Reference Center For Huntington's Disease

| Title | Funding source |
|---|---|
| Neuro HD : Huntington's disease and neuroleptics - Comparisons of olanzapine , tetrabenazine and tiapride . A controlled multicenter , randomized, study | APHP, PHRC P060211 |
| A multi-centre, multi-national prospective observational imaging biomarker study in early stage Huntington's disease (HD) patients to assess imaging techniques and parameters able to support efficacy studies with SEN0014196 in HD patients during Phase II and III studies | FP7 |
| REGISTRY An observational study of the European Huntington Disease | EHDN |
| Multicentre trial of treatment of Huntington's disease by cysteamine ( CYST HD) | PHRC2004- 03bis, DRC Angers . |
| Imalang : language processing in HD | ANR |
| Extension of the multicentric intracerebral grafting trial in HD : Post-MIG-HD | PHRC |
| Repair HD : toward stem cell transplantation in HD | FP7 |
| Radico cohort | French investement for the future |
| EcogHD ; e-cohort in Huntington's disease | AO CRC DRCD |
| Mettre les projets de la Salpétrière | |

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| PRIDE HD | TEVA |

Universitätsklinikum Bonn

| Title | Funding source |
|---|---|
| Langzeitbeobachtung dementieller Symptome und cognitiver Parameter sowie Anwendbarkeit neuer prognostischer Marker bei der Parkinson-Erkrankung (LANDSCAPE) | BMBF |
| Biomarkers for Alzheimer´s disease and Parkinson´s disease (BIOMARKAPD) | JPND |
| Integrated European Project on Omics Research of Rare Neuromuscular and Neurodegenerative Diseases (Neuromics) | FP7 |
| Patienten ohne Diagnose: Aufbau einer Interdisziplinären Kompetenzeinheit zur Erkennung Seltener chronischer Erkrankungen | Robert-Bosch-Stiftung |
| IDSEM-Verbundprojekt. Integrative Datensemantik für Neurodegenerative Forschung, Teilprojekt C | BMBF |
| European Spinocerebellar Ataxia Type 3/Machado-Joseph Disease Initiative (ESMI) | JPND |

Institute of Neurogenetics, University of Lübeck

| Title | Funding source |
|---|---|
| Project C5 “Connectivity and plasticity in cortical motor networks in Parkin gene associated parkinsonism and dopa responsive dystonia” as part of the SFB 936 “Multi-Site Communication in the Brain” | DFG, SFB 936 |
| Dystract consortium – translational research of dystonia | BMBF |
| Project C5 “Modulation of the action selection and error processing networks in genetic parkinsonism using rTMS and DBS“ as part of the SFB 936 (second funding period) | DFG, SFB 936 |
| Project grant "New diagnostic approach to patients with rare diseases | UKSH Förderstiftung |
| „Characterization of endophenotypes in focal task-specific dystonia” | Dystonia Coalition, NIH |
| „Transcanial ultrasonographic evaluation of the basal ganglia in DYT1, DYT11 and DYT12 dystonia“ | The Bachmann-Strauss Dystonia & Parkinson Foundation |
| „The role of endogenous Parkin and PINK1 mutations in human dopaminergic neurons”; KL1134/11-1 | Deutsche Forschungsgemeinschaft |
| „StemBANCC; IMI Project on human induced pluripotent stem cells” | EC (FP7) |
| “Comprehensive Unbiased Risk factor Assessment for Genetics and Environment in Parkinson´s Disease” | BMBF |
| „MitoPD Mitochondrial endophenotypes of PD” 031A430B | BMBF |
| „Molekulare Charakterisierung von TUBB4-Mutationen”; KL1134/13-1 | DFG |
| „Genetic risk factors of musician´s dystonia“; LO 1555/4-1 | DFG |
| „Elucidation of the genetic cause of dystonia 4 using exome sequencing”; | Bachmann Strauss Dystonia & Parkinson Foundation |
| „Medizinische Genetik – Von seltenen Varianten zur Krankheitsentstehung“; | University of Lübeck |
| „Identification of targets and interactors of the DYT6-related transcription factor THAP1“; LO 1555/3-2 | DFG |
| “Unraveling novel genetic cause in alcohol-responsive dystonia by exome sequencing” (NS065701) | Dystonia Coalition (NIH-funded) |
| „Identifizierung neuer Dystonie-Gene bei konsanguinen Familien“; LO 1555/8-1 | DFG |
| Systems Medicine of Mitochondrial Parkinson’s Disease | European Union’s Horizon |

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| SYSMED-PD (http://sysmedpd.eu/) | 2020 research and innovation programme under grant agreement No 668738. |
| mineRARE - Semantic text-mining methods to screen for rare disorders / Semantisches Textmining zur Identifikation seltener Erkrankungen (mineRARE) | In collaboration with the Friedrich-Baur-Institute, LMU München, external funding |

Klinikum der Ludwig- Maximilians-Universität München

| Title | Funding source |
|---|---|
| TIRCON (Treat Iron-Related Childhood-Onset Neurodegeneration) (as Coordinator) | EU-FP7 |
| EFACTS (European Friedreich's Ataxia Consortium for Translational Studies) (as Co-Investigator) | EU-FP7 |
| DESCRIBE (DZNE – Clinical Register Study of Neurodegenerative Disorders) (as Co-Investigator) | BMBF-DZNE |
| EOA (Early Onset Ataxias) (as Co-Investigator) | BMBF-DZNE |
| HSP-Net (Vernetzungsprojekt Hereditäre Spastische Spinalparalyse) (as Co-Investigator) | BMBF-DZNE |
| ALCAT (Effects of acetyl-DL-leucine on cerebellar ataxia trial: a multicenter, multinational, randomized, double-blind, placebo-controlled, 2-way, crossover phase III trial) (as Co-Inv.) | BMBF |

University Hospitals Leuven

| Title | Funding source |
|---|---|
| 'The contribution of the medial temporal system in humans to semantic processing of words and pictures' (G092515N), PI (280,000 Euro) (PI) | Flemish Research Foundation (FWO) |
| May 1, 2014 - April 30, 2018: Vlaamse Impulsfinanciering voor Netwerken voor Dementie-onderzoek (2.5 million Euro, KU Leuven coordinator Rik Vandenberghe | Agency for Research and Innovation (IWT) |
| Jan 2013 - Dec 2017: Strategisch Basisonderzoek agentschap voor Innovatie door Wetenschap en Technologie 'Biomarker based adaptive development in Alzheimer Disease (BioAdaptAD)' (120835), projectleider module 2 (totaal 979.720,17 Euro) | Agency for Research and Innovation (IWT) |
| 2012-2014: Partner Hercules Large-scale Research Infrastructure 'An integrated multimodal platform for brain mapping at high spatiotemporal resolution: 3 Tesla MRI infrastructure' | Hercules Foundation |
| October 1, 2012 - September 30, 2016: Bijzondere Onderzoekstoelage OT/12/097: Functional segregation and integration in the cortical circuits for language and selective attention and the effects of focal lesions and cortical neurodegenerative disease. Principal Investigator (total of 450,000 Euro). | KU Leuven |
| January 1, 2013 – December 15, 2015: Towards an integrated drug discovery platform for tau pathogenesis | IWT |
| January 1, 2009 - December 31, 2012: FWO G.0660.09N Het bihemisferisch netwerk voor taal en semantisch<br>geheugen in de intacte hersenen, bij corticale neurodegeneratie en bij corticale ischemie, Promotor Rik Vandenberghe (total of 218,315 Euro) | FWO |
| 18F-THK5351 tau PET imaging in Alzheimer's disease, frontotemporal degeneration and healthy older adults (150,000 Euro) | Stichting Alzheimer Onderzoek |
| 2012 – 2016: 'PET imaging of Phosphodiesterase 10A (PDE10A) in Huntington's disease, Progressive supranuclear palsy and Parkinson's disease'. Co-investigator | KU Leuven |

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| Wim Vandenberghe. | |
| 2013-2016: 'Mitochondrial imaging in live neurons from Parkinson's disease patients' (G.0A44.13). Co-investigator Wim Vandenberghe. | Flemish Research Foundation (FWO) |
| 2012-2017: 'Parkinson's disease: the mitochondrial connection' (Geconcerteerde Onderzoeksactie GOA/13/017). Principal investigator Wim Vandenberghe (total amount: 900.000 €) | KU Leuven |
| 2012-2015: 'The role of deubiquitinases in Parkin-mediated mitophagy and Parkinson's disease' (G.0583.12N). Principal investigator Wim Vandenberghe (total amount: 296.000 €) | Flemish Research Foundation (FWO) |
| 2011-2013: 'Motor learning in Parkinson's disease: A randomized comparison of dual and single task gait practice'. Co-investigator Wim Vandenberghe. | Jacques and Gloria Gossweiler Foundation |
| 2011 - 2014: 'Neuroplasticity in Parkinson's Disease: consolidation of motor learning and its related brain activity changes' (G.0906.11). Co-investigator Wim Vandenberghe. | Flemish Research Foundation (FWO) |
| 2010-2013: 'In vivo study of the functional role for type 1 and 2 cannabinoid receptors in neurodegeneration' (G.0493.10N). Co-investigator Wim Vandenberghe. | Flemish Research Foundation (FWO) |
| 2008-2011: 'Analysis of the composition and function of the parkin complex' (G.0607.08). Principal investigator Wim Vandenberghe (total amount: 84.000 €). | Flemish Research Foundation (FWO) |
| 2015-2017: '"Parkinson's Disease: Mechanisms, Biomarkers and Therapeutics". Principal investigator Wim Vandenberghe (total amount; 200.000 €). | Campaign Opening the Future (KU Leuven) |

Istituto Neurologico Carlo Besta, Milan

| Title | Funding source |
|---|---|
| Monitoring disease progression and phenotypic heterogeneity in hereditary ataxias: clinical, cognitive, neuroimaging, and neurophysiological study to identify early markers of cerebellar dysfunction in patients and presymptomatic carriers ( | Italian Ministry of Health, Grant RF-2011-02347420 (CM) |
| An Italian study on intermediate-lenght polyQ-tract expansions: frequency, clinical variability, and brain morphometry of subjects at risk for late-onset neurodegenerative diseases | Italian Ministry of Health, Grant GR-2013-02357821 |
| FP7 European Union Project "Leuko-Treat" | European Community-HEALTH-F2-2010-241622 |
| TIRCON: Treat Iron Related Childhood Onset Neurodegeneration | European Community-(FP7/2007-2013, HEALTH-F2-2011, grant no. 277984) |
| GENetic Frontotemporal dementia Initiative (GENFI I) | Centres of Excellence in Neurodegeneration Grant (2012-2014) |
| GENetic Frontotemporal dementia Initiative (GENFI II) | Centres of Excellence in Neurodegeneration Grant (2015-ongoing) |
| Joint Programming Neurodegenerative Diseases:<br>Searching for therapeutic interventions in frontotemporal dementia with C9orf72 repeat expansions in the presymptomatic stage | 2015-ongoing |
| Phenotypic characterization of parkinsonian syndromes associated with glucosecerebrosidase mutations (GR-2009-1607326) | Italian Ministry of Health, Grant GR-2009-1607326 |
| DBS tourrette Subthalamic Nucleus deep brain stimulation in Tourette's syndrome (GR-2009-1594645) | Italian Ministry of Health, Grant GR- |

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| | 2009-1594645 |

Bambino Gesù Children's Research, Rome

| Title | Funding source |
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| DRUG REPOSITIONING IN RHO-LINKED COGNITIVE IMPAIRMENT: FASUDIL AND OTHER ROCK INHIBITORS AS POTENTIAL TREATMENTS IN OPHN1 SYNDROME | Mariani Foundation |
| Identification of genes involved in hypomyelinating white matter disorders 2011 | ELA Foundation |

VU University Medical Center, Amsterdam

| Title | Funding source |
|---|---|
| Metachromatic leukodystrophy: Gallbladder abnormalities and their clinical relevance | Private gifts |
| Genetic causes of leukodystrophies | ZonMw TOP |
| Metachromatic leukodystrophies: Quantitative MRI approaches | Metakids |
| Vanishing white matter: natural history | |

University Medical Center Groningen

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| Actelion research foundation:<br>Screening for Nieman Pieck type C among patients with dystonia; a systems diagnostics approach. 2016 €50,000 (de Koning) |
| Metabolic power foundation:<br>"Unraveling psychiatric co-morbidity in dopa-responsive dystonia: is serotonin the common pathway?" 2016 €19,389 (de Koning) |
| Phelps Stichting:<br>Moving forward in early onset dystonia<br>2015 100.000 euro (de Koning-Tijssen) |
| Jacques and Gloria Gossweiler Foundation & Fonds Nuts-Ohra:<br>Effectiveness of a standardized physical therapy program in Cervical Dystonia; a single blind randomized controlled trial.<br>2014 200.000 euro (de Koning-Tijssen) |
| ZonMW Netherlands organisation for Scientific Research, program Doelmatigheidsonderzoek Farmacotherapie Review dure geneesmiddelen.<br>Botulinum toxintreatment in focal dystonia.<br>2010-2011 48.000 euro (de Koning-Tijssen) |
| PBF Prinses Beatrix Fonds & Stichting wetenschapsfonds dystonie vereniging<br>DystonieNet : development of botulinum toxin evidence based guideline for cervical dystonia.<br>2010 120.000 euro (de Koning-Tijssen) |
| STW Technology society, program: Perspective - NeuroSIPE 2009.<br>One of principal investigators project: Torticollis: Identification of cortical and spinal motor contributions in cervical dystonia<br>2010 1.000.000 euro (de Koning-Tijssen) |
| PBF Prinses Beatrix Fonds<br>'Genetic associations in dystonia'<br>2010 240.000 euro (de Koning-Tijssen) |
| Metakids research grant: |

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| Movement disorders in children with inborn errors of metabolism.<br>2013 75.000 euro (de Koning) |
| EHDN Enroll funding (huntington's disease)<br>2014 60.000 euro (Kremer) |
| Prinses Beatrix Fonds<br>The spinocerebellar ataxias: from gene identification to protein function<br>2011 € 250.000 (Verbeek) |
| Hersenstichting Nederland (Netherlands brain foundation)<br>Functional characterization of non-classified variations in spinocerebellar ataxia type 13<br>2011 € 35,000 (Verbeek) |
| NutsOhra Fund<br>Whole exome sequencing: the route to maximal diagnostics for ataxia patients 2011 € 42,500 (Verbeek) |
| eRARE young investigator grant Towards the Understanding of Pathological Protein Processing and Toxicity in Machado-Joseph Disease<br>2013 €139,500 (Verbeek) |
| EU KP7 Health<br>Treating Iron Related Childhood Onset Neurodegeneration (TIRCON)<br>2011 € 5,200,000 (Sibon, full partner and workpackage leader) |
| EU E-rare project<br>European multidiciplinary initiative on neuroacanthocytosis (EMINA)<br>600.000 euro<br>2010(Sibon, full partner) |
| EU KP7 Health<br>Treating Iron Related Childhood Onset Neurodegeneration (TIRCON)<br>2011 € 5,200,000(Sibon, full partner and workpackage leader) |

Radboud University Medical Center, Nijmegen, The Netherlands

| Title | Funding source |
|---|---|
| Cerebellar dysfunction and its compensation in presymptomatic carriers of dominant ataxia genes: heading towards therapeutic neuromodulatory interventions in ataxias. | Gossweiler Foundation |
| Physiotherapy in ataxia: effectivess and usage | KNGF |
| Cerebral compensatory mechanism in spinocerebellar ataxias | Radboudumc |
| Systematic Modeling of Autosomal Recessive Cerebellar Ataxias in *Drosophila* - novel avenues towards better understanding and future treatment | Radboudumc |
| Phenotypic enrichment of the Nijmegen Parkinson's disease genetic database | BBMRI-NL |
| The effect of cerebellar cTBS on writer's cramp | Dutch Dystonia Society |
| European Spinocerebellar Ataxia Type 3/Machado-Joseph Disease Initiative | EU/ZonMW |
| Preparing for therapies in autosomal recessive ataxias | EU/ZonMW |
| Understanding balance problems through studying startle reactions in hereditary spastic paraplegia | Radboudumc |
| Sjogren-Larsson Syndrome: clinical studies | Radboudumc |
| GLUT1 deficiency syndrome: clinical aspects, treatment, genetics, and biomarkers | ZonMW |
| Ataxia telangiectasia: genetics in relation to clinical aspects | Stichting AT |
| Ataxia telangiectasia: movement disorders; prognosis; consequences of carriership | Stichting AT |

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| | Twan Foundation |

University Medical Centre Ljublijana

| Title | Funding source |
|---|---|
| Identification of new disease mechanisms with next generation sequencing (B. Peterlin) | ARRS |
| Integrative genomics in neurodegenerative diseases (B. Peterlin) | ARRS |
| Early detection and rational therapy of dementia: role of biomarkers (Z. Pirtosek) | ARRS |
| Targeting new receptors in dystonia: electrophysiological and neuroimaging correlates of the effect of zolpidem, a selective agonist of benzodiazepine subtype receptor alfa-1, in different forms of primary focal dystonia (M. Kojovic) | ARRS |
| Cardio-respiratory responses during hypoxic exercise in individuals born prematurely (D. Osredkar) | ARRS |

Pitié-Salpêtrière hospital, Paris, France

Selected projects among 25 projects (period 2011-2016)

| Title | Funding source |
|---|---|
| FTDGenes.<br>Constitution d'une cohorte clinico-génétique nationale de patients pour identifier de nouveaux gènes impliqués dans les formes monogéniques de DLFT'.<br>2008-2011. | ANR (programm MRARES 2008)<br>300 000 € |
| Predict-PGRN.<br>Caractérisation multimodale des individus porteurs de mutation du gène PGRN à un stade symptomatique ou présymptomatique de la maladie.<br>2010-2019 | Programme Hospitalier de Recherche Clinique<br>& Fondation Plan Alzheimer<br>500 000 € |
| Modélisation du phénotype et de la réversion de la DFT liée à la progranuline.<br>2010-2013 | Fondation-plan Alzheimer. AAP Animal models<br>350 000 € |
| Typage-APP/CAPP.<br>Caractérisation linguistique, anatomique/métabolique et biologique des différentes formes d'aphasie primaires progressives: vers un rationnel pour des rééducations ciblées et des essais pharmacologiques.<br>2010-2016 | Programme Hospitalier de Recherche Clinique (PHRC) national 2010<br>740 000 € |
| RiMOD –<br>2013-2016 | JPND<br>300 000 € |
| FTLD-exome<br>2015-2019 | Programme Hospitalier de Recherche Clinique (PHRC) national<br>700 000 € |
| NeurOmics<br>Integrated European Project on Omics Research of Rare Neuromuscular and Neurodegenerative Diseases – | FP7-Seventh Framework Programme for Research and |

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| 2013-2017 | Technological Development<br>300 000 € |
| PrevDemAIs<br>Predict to prevent FTD and ALS<br>2015-2019 | ANR –PRTS<br>1 700 000 € |
| InCure<br>Innate Immune Activation in Neurodegenerative Disease.<br>2014-2017 | JPND<br>200 000 € |

Vall Hebron University Hospital

| Title | Funding source |
|---|---|
| Diagnostic exome, research exome and whole genome: maximising NGS performance in early infantile epileptic encephalopathies (PI15/ 01791) PI: A.Macaya | Instituto de Salud Carlos III (Spanish health Ministry) |
| Whole Exome Sequencing for the molecular analysis of early infantile epileptic encephalopathies (PI12/1005) PI: A. Macaya | Instituto de Salud Carlos III (Spanish health Ministry) |
| Exome sequencing of early epileptic encephalopaties: data analysis (2010 BP_A2 00025) PI: A.Macaya | AGAUR –Beatriu de Pinos grant |
| Eurohead Pain. Genetics work package. PIs (Barcelona node): A.Macaya/P.Pozo | European Union FP 7th Framework programme |
| Genes and migraine:Positional cloning in familial migraine and expression in experimental cortical spreading depression. (SAF2009-13182-C03-03) PI: A.Macaya | MICINN (Science & Innovation Spanish Ministry) |
| Investigation of the of HLA-DRB1/DQB1 polymorphisms as susceptibility genetic factors in sporadic and familial forms of autoimmune myasthenia gravis in a Spanish population. PI: J.Gámez | Instituto de Salud Carlos III (Spanish health Ministry) |
| DESATA: a project for development of a drug for treating transthyretin amyloidosis. PI: J.Gámez | Spanish Economy and Competitivity Mnistry |
| Structural lesions and neuropsychological repercussions in mild traumatic brain injury (PI13/02397) | Fondo Investigación Sanitaria, (Instituto de Salud Carlos III) co-financed by the European Regional Development Fund |
| Molecular mechanisms involved in the genesis of the glioma and study of the tumoral stem cells. Identification of new therapeutic targets and markers for the stratification in patients and response to drugs (AECC-GE2010-07) | Asociación Española contra el Cancer |
| Neuropsychological impairment and brain lesions in mild traumatic brain injury (MAPFRE 2012-04) | Fundación Mapfre |
| Analysis of the temporal profile of the neuroinflammatory response mediators in the brain extracellular space by high-resolution microdialysis after severe traumatic brain injury (FMM-2010-10) | Fundación Mútua Madrileña |
| Applications of high-resolution microdialysis to define the molecular profile of post-traumatic and ischemic brain edema and the contribution of non-selective SUR1-regulated NCCa-ATP channels to its formation. (PI10/00302) | Fondo Investigación Sanitaria, (Instituto de Salud Carlos III) co-financed by the European Regional Development Fund |
| Analysis of the metabolic and inflammatory effects of cortical spreading depression (CSD) and CSD-like phenomena in acute brain lesions (PI080480) | Fondo Investigación Sanitaria, (Instituto de Salud |

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| | Carlos III) co-financed by the European Regional Development Fund |
| Analysis of the temporal profile of the neuroinflammatory response mediators in the brain extracellular space by high-resolution microdialysis after severe traumatic brain injury (MAPFRE 2009/01) | Fundación Mapfre Medicina |
| Advanced Arterial Hypotension Adverse Event prediction through a Novel Bayesian Neural Network (AVERT-IT) (FP7-217049) | European Commission. Seventh Framework Programme |
| Role of the neuropeptides hypocretin-1, melatonin and cortistatin in sleep-wake cycle alterations in patients with normal pressure hydrocephalus (PI070681) | Fondo Investigación Sanitaria, (Instituto de Salud Carlos III) co-financed by the European Regional Development Fund |