Research calls and collaborative Genetic Reseach

Dear all,

As there are a few **new research calls** and 4 new calls for **collaborative genetic research**, here is an update!

You can read more information about the new funding opportunities on the webpage of the research council. **The password to access the page is: 0124.**

Regarding the collaborative research calls, this is the webpage.

Calls for collaborative genetic research

Adult phenotype of CDKL5 deficiency disorder

Targeted gene(s)/phenotype under study: CDKL5

Summary: Pathogenic variants in the cyclin-dependent kinase-like 5 (CDKL5, Xp22.13) gene are the cause of CDKL5 deficiency disorder (CDD) and one of the main etiologies of genetic epilepsy, with an estimated birth prevalence around 1 in 40,000. While the phenotype and evolution of the CDD-related developmental and epileptic encephalopathy (DEE) is well characterized during the first years of life, the adult phenotype is not yet fully understood. It is known that other genetic DEEs show specific adult phenotypes, such as parkinsonism in Dravet syndrome or psychotic features in PCDH19-DEE, apart from the epilepsy evolution. The aim of this project is to characterize the adult phenotype of patients with CDD. Comprehensive clinical data, as well as EEG raw data, will be collected and evaluated.

Coordinating clinicians: Angel Aledo-Serrano and Rikke Moller from the Epilepsy Genetics dpt, Danish Epilepsy Center, Dianalund, Danemark

Genetic and clinical characterization of patients with microdeletions of KCNQ2 and EEF1A2

Targeted gene(s)/phenotype under study: KCNQ2 + EEF1A2

Summary: We aim at describing the clinical (epilepsy, neurodevelopmental,…) and EEG phenotype of patients with a microdeletion (CNV loss) including both KCNQ2 and EEF1A2 We need precise CNV coordinates, family history and parental genetic testing if available.

Coordinating clinicians: Gaëtan Lesca, Alice Bergevin and Clotilde Rivier from the medical genetics dpt, University Hospital of Lyon, France

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Genetic and environmental modifiers associated with KCNQ2-related disorder

Targeted gene(s)/phenotype under study: KCNQ2 (OMIM: 613720 and 121200; ORPHA: 439218)

Summary: KCNQ2 encodes for a voltage gated potassium channel subunit that has a critical role in controlling neuronal excitability. KCNQ2-related disorders are associated with a spectrum of phenotypes ranging from self-limiting (familial) neonatal epilepsy at the mild end to developmental and epileptic encephalopathy at the severe end. Neonatal seizures are the main features, but patients without neonatal seizures are described. Although phenotype-genotype correlations are good, phenotypic variability exists even among carriers of recurrent variants.
We hypothesize that some of the phenotypic variability in KCNQ2-related disorders can be explained by the existence of genetic and environmental modifiers. By identifying these modifiers, we hope to improve counseling for individual patients, and to identify biological pathways that can be targeted by novel disease-modifying treatment strategies.
In this study, all patients with a proven pathogenic KCNQ2 variant can be included after signature of an informed consent document. Detailed clinical data on all participants will be collected through the treating (child) neurologist, in addition to a DNA aliquot.

Coordinating clinician: Sarah Weckhuysen from the Department of neurology, University Hospital of Antwerp, Antwerp, Belgium

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SLC35A2-related epilepsy: specific electroclinical feature

Targeted gene(s)/phenotype under study: SLC35A2

Summary: Pathogenic de novo variants in the X-linked gene SLC35A2 encoding the major Golgi-localized UDP-galactose transporter cause a rare type of congenital disorder of glycosylation known as SLC35A2-congenital disorders of glycosylation (CDG). In addition, somatic mutations in this gene has been implicated in pathogenesis of some types of malformations of cortical development (Mild malformation of cortical development with oligodendroglial hyperplasia and epilepsy - MOGHE). Patients with germline mutations in SLC35A2 present a severe neurodevelopmental disorser, frequently associated with epilepsy and different degrees of systemic comorbidities, and galactose supplementation might be useful for their treatment. However, the specific electroclinical syndrome of this patient has

not specificly establish yet and studies on this topic are scarce. The aim of this project is to characterize the specific epilepsy phenotype of patients with mutations in SLC35A2, their differences with patients with MOGHE, and their response to treatments, including galactose supplementation. Comprehensive clinical data, as well as EEG raw data, will be collected and evaluated.

Coordinating clinician: Angel Aledo-Serrano and Rikke Moller from the Epilepsy Genetics dpt, Danish Epilepsy Center, Dianalund, Danemark.

Calls for collaborative genetic research

Research Calls

1. EJPRD: Joint Transnational Call 2022

The aim of the call is to enable scientists in different countries to build an effective collaboration on a common interdisciplinary research project based on complementarities and sharing of expertise, with expected impact to use the results in the future for benefit of patients.

Deadline model: two-stages
Deadline date: 16th February 2022; 15th June 2022
Grant duration: max. 3 years

More information

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2. EJPRD: Networking Support Scheme (NSS) call

The **first aim** of the Networking Support Scheme in the European Joint Programme on Rare Diseases (EJP RD) is to **encourage sharing of knowledge on rare diseases or rare cancers between health care professionals, researchers and patients in new or expanding research networks by funding networking events.**
The **second aim** of the Networking Support Scheme is to enable or increase the participation of usually underrepresented countries in Europe in new and in expanding research networks on rare diseases or rare cancers.

Deadline model: continuous basis; Events may be organized between 6 and 18 months after the application date for 2021. From 2022 onwards, events may be organized between 6 and 12 months after the application date.
Budget: max. 30.000€ per event

More information

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3. Sergievsky Award for Epilepsy Health Equity and Diversity

The **Sergievsky Award for Epilepsy Health Equity and Diversity** is a brand-new AES research award for early-career physicians and scientists who identify as members of underrepresented racial and ethnic groups, with preference for those who are Black or African American, and/or who are researching issues that affect medically underserved individuals with epilepsy or seizures or related aspects of health equity.

Deadline date: Friday, January 14, 2022
Budget: $150,000 over two years

More information

More information on these calls - code 0124

European Commission

European Reference Networks

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Our mailing address is:

Department of Paediatric Clinical Epileptology, Sleep Disorders and Functional Neurology
ERN EpiCARE c/o Pr Arzimanoglou
Hôpital Femme Mère Enfant (HFME)
59 Boulevard Pinel
69677 Bron, FRANCE