



**DESIRE**  
Development & Epilepsy



Strategies for Innovative Research to  
Improve Diagnosis, Prevention and Treatment  
In Children with Difficult to Treat Epilepsy

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Many childhood epilepsies, especially severe forms, result from a complex combination of  
**an epileptogenic process  
and a developmental process,**  
which are thought to influence each other.

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# Why EED?

- A major cause of **EED** are **malformations of cortical development (MCD)**, either macroscopic or subtle.
- **EED** are often manifested as **epileptic encephalopathies (EE)**, i.e. conditions in which epileptic activity itself may contribute to severe cognitive and behavioral impairments.
- **EED** are the most frequent drug-resistant pediatric epilepsies carrying a lifelong perspective of disability and reduced quality of life.
- Although **EED** collectively represent a major medical and socio-economic burden, their molecular diagnosis, **pathogenic mechanisms (PM)** and rationale treatment are poorly understood.



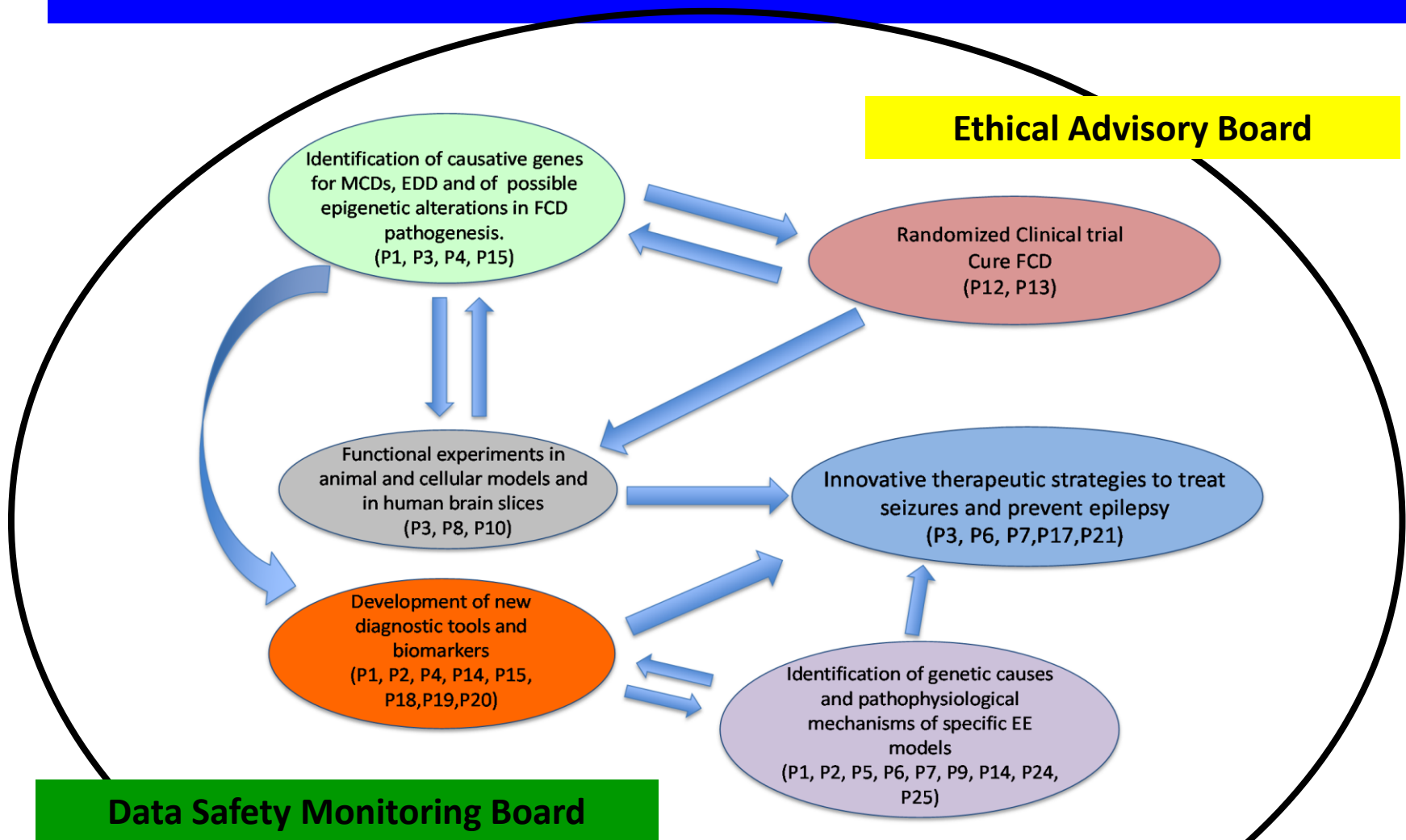
# What are the objectives of DESIRE?

To advance the state of the art with respect to:

- the genetic and epigenetic causes and **PM of EDD, particularly epileptogenic MCD**
- to elucidate **molecular networks and disrupted protein complexes** and search for common bases for these apparently heterogeneous disorders.
- To develop and use **diagnostic tools (biomarkers)** and protocols through the study of a unique and well-characterized cohort of children to provide standardized diagnosis for patient stratification and research across Europe.
- To develop **treatments for EDD** using randomized, multidisciplinary clinical protocols and test preclinical strategies in experimental models which address novel preventative strategies.



# How do we collaborate?



# DESIRE in numbers

- 5 years project starting the 1<sup>st</sup> October 2013 – Last year 2018
- 28 partners (last submitted amendment)
- 8 Third Parties
- 9 countries
- 2.227,29 man/months
- > 250 researchers



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# Who are we?

UNIVERSITA DEGLI STUDI DI FIRENZE (co-ordinator)

FONDAZIONE IRCCS ISTITUTO NEUROLOGICO CARLO BESTA

INSTITUT NATIONAL DE LA SANTE ET DE LA RECHERCHE MEDICALE (INSERM)

UNIVERSITAETSKLINIKUM ERLANGEN

UNIVERSITA DEGLI STUDI DI VERONA

CENTRE NATIONAL DE LA RECHERCHE SCIENTIFIQUE

ISTITUTO ITALIANO DI TECNOLOGIA

AGENCIA ESTATAL CONSEJO SUPERIOR DE INVESTIGACIONES CIENTIFICAS

KING'S COLLEGE LONDON

CHARITE - UNIVERSITAETSMEDIZIN BERLIN

KLINIKUM DER UNIVERSITAET ZU KOELN

UNIVERSITY COLLEGE LONDON

THE UNIVERSITY OF LIVERPOOL

UNIVERSITE LIBRE DE BRUXELLES

CHRISTIAN-ALBRECHTS-UNIVERSITAET ZU KIEL

BAKER IDI HEART AND DIABETES INSTITUTE HOLDINGS LIMITED

UNIVERSITA TA MALTA

UNIVERSITA TA MALTA

DI.V.A.L. TOSCANA SRL

MICROMED S.P.A.

VARIONOSTIC GMBH

CEGAT GMBH

AMARNA THERAPEUTICS BV

CF CONSULTING FINANZIAMENTI UNIONE EUROPEA SRL

UNIVERSITA CATTOLICA DEL SACRO CUORE

OSPEDALE PEDIATRICO BAMBINO GESU

ISTITUTO GIANNINA GASLINI

COMMISSARIAT A L ENERGIE ATOMIQUE ET AUX ENERGIES ALTERNATIVES

UNIVERSITE D'ANGERS

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Thank you!



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