The pleiotropic effects of ADNP in Mental Disorders (ADNPinMED)

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Despite the unprecedented number of recent novel disease gene identifications in neurodevelopmental disorders such as intellectual disability, autism, or schizophrenia, our understanding of the pathogenicity mechanisms and associated clinical spectra are limited. We are an existing, established and productive ERA-NET neuron consortium. In our ongoing network, we studied mutations in ADNP, originally identified as a gene involved in syndromic autism, using a suite of cellular and animal models and tools developed. Since our results obtained in the ongoing project indicated a much broader clinical phenotype than anticipated and linking ADNP to multiple dimensions of epigenetic regulation, we now propose to apply our resources to investigate the involvement of the epigenome in the phenotypical presentation of mental disorders, using ADNP as a model.
Our work will be based on the materials we generated in the ongoing application, including unique cellular and specifically for this project created animal models of the disorder. The work plan consists of six work packages, including disease characterization in patients and animal models, transcriptomics and epigenomics, functional analysis, mosaicism analysis, data integration and preclinical drug testing. These results will enable a full characterization of the consequences of the ADNP mutation that can be linked to the specific aspects of the diseases caused by ADNP mutations.