https://genida.unistra.fr
Updated August 2018

Context of the GENIDA project

Current status of genetic forms of Intellectual disability (ID), autism spectrum disorders (ASD) & epilepsy

- **Intellectual disability**, with or without **autism** manifestations or **seizures**, affects about **1.5 to 2%** of children or young adults (~0.4-0.5% for the more severe forms). Through use of exome or genome sequencing, it is estimated that 50-60% of cases are due to **single genetic causes** (single gene mutation or copy number variation - CNV). It is a huge **public health and social problem**.

- Many **recurrent CNVs** and **more than 700 known genes*** are implicated in genetic forms of intellectual disability (ID) and autism spectrum disorders (ASD) (and are often associated to epilepsy), and thus define as many rare diseases. In most cases these genes and CNVs have been recently identified (and novel genes are added to the list continuously). There is in general **very limited information on the clinical spectrum, comorbidities** and especially on the **natural history** (evolution of the disease over life).

  ** L. E. L. M. Vissers, C. Gilissen and J. A. Veltman - Nat Rev Genet 2016 (doi:10.1038/nrg3999)*

- How can we **efficiently build cohorts of patients** (in the context of individually rare or very rare diseases – ie international), and obtain sufficient and thorough data to identify information of medical interest for both families and professionals?

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The GENIDA project

A participatory international database to collect medically relevant information on genetic forms of ID/ASD, for families and professionals

- Collect longitudinal health data on **genetic forms** of Intellectual Disability (ID), Autism Spectrum Disorders (ASD) and epilepsies associated to ID through the **direct Internet participation of families** (*https://genida.unistra.fr).* Families answer to a structured multiple choice **questionnaire** and five additional « open answer questions » concerning major health and quality of life problems.
- Allow **participating families** and **professionals** to access to our **de-identified statistical analyses,** a feature already activated for the five largest cohorts.
  - **Empower patients/families** by allowing them to directly participate to the project and giving them feedback on the health characteristics caused by the specific gene defect found in their child.
- Generate, for specific genetic forms of ID/ASD, **novel and medically significant knowledge** that can be translated in improved personalized healthcare.
- Be a **platform for professionals** who wish to submit **additional specific questions** to subset of patients or even recruit patients for ethically approved research or clinical studies. Such proposals would be screened by the GENIDA Advisory Committee.