Genetics of Intellectual Disability and Autism Spectrum Disorders

GenIDA is an **international participatory database** that collects medical information from families **to better characterize the clinical manifestations and natural histories of genetic forms of intellectual disability (ID) and/or autism spectrum disorders (ASD).**

Context

• **Intellectual disability**, with or without manifestations of **ASD** and/or **epilepsy**, affects **1-2% of the population** (approx. 0.4-0.5% for the most severe forms).
Thanks to the use of exome and genome sequencing, it is estimated that over 50% of these cases have a **single genetic cause** (mutation in a gene, chromosomal abnormality or copy number variation). This represents a **major public health problem**.

• More than **1,000 genes and recurrent chromosomal abnormalities** are involved in these genetic forms of ID or ASD (frequently associated with epilepsy), which constitute so many different rare diseases that often remain **insufficiently described in terms of clinical spectrum, associated medical problems and natural history** (evolution of the pathology over the course of a lifetime), due to their rarity and the often limited number of patients observed.

• In this context of (very) rare pathologies, how can we **efficiently build patient cohorts** and obtain sufficient quality data to identify new information of medical interest to families and professionals?

The project

- **Collect longitudinal health information on genetic forms of ID**, whether or not associated with ASD and/or epilepsy, through the **direct participation of families** who answer a **structured questionnaire accessible online** (https://genida.unistra.fr).
- **Involve families** by enabling them to participate directly in the project, and by informing them of the health characteristics specific to genetic anomalies affecting their relatives.
- Give **participating families** and **medical and paramedical professionals** access to our **de-identified statistical analyses**.
- Produce, for each specific form of ID/ASD, **new medically significant knowledge** that can be translated into improved patient care.
- **Provide a platform for professionals** to **submit additional questions** to patient cohorts, or to **recruit patients for ethically approved clinical studies and/or research projects** (subject to approval by GenIDA's Scientific Advisory Board).