

# Italian GLUT1DS registry: preliminary report after one year of running

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**Background:** Patient registries are rapidly growing to implement research. Registries are powerful tools providing insights and real-world data about the epidemiology, phenotypic spectrum, diagnostic biomarkers, effectiveness of treatments, and opportunities for quality improvement of healthcare delivery. This report describes the rationale, methods and initial implementation of the Italian GLUT1DS registry

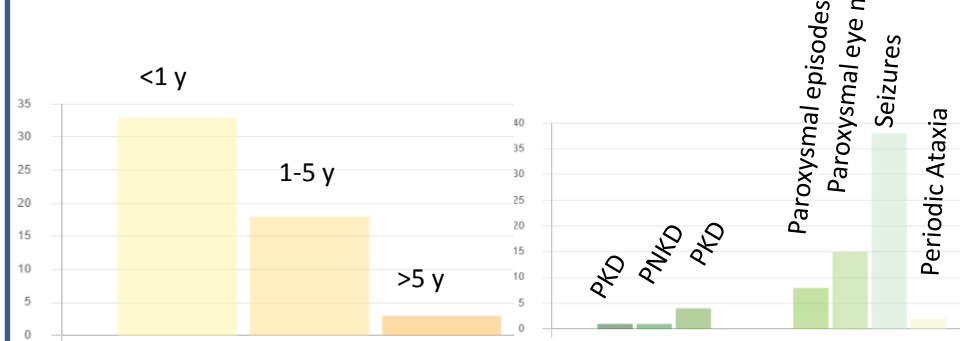
**Methods:** The Italian GLUT1DS registry is an ongoing retrospective and prospective, multicenter, observational registry, developed in collaboration with the Italian GLUT1DS association. It is based on an informatics flexible technology platform (cloud-R), structured according to the most rigorous legal national and European requirements for management of patient's sensitive data. It collects baseline and follow-up data on the patient's demographics, history, symptoms, genotype, clinical and instrumental evaluations, therapies.

## Participating centers

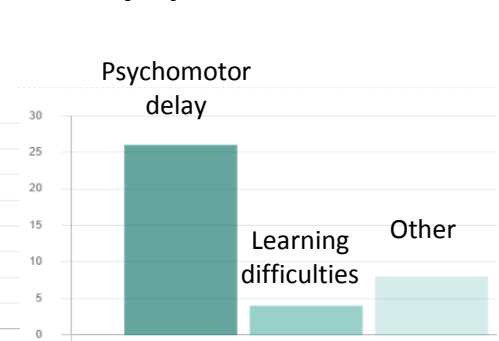
IRCCS Mondino, Pavia	23
Ospedale Vittore Buzzi, Milano	18
Ospedale Meyer, Firenze	11
Istituto Neurologico Besta, Milano	4
Ospedale Gaslini, Genova	3

## Preliminary Results

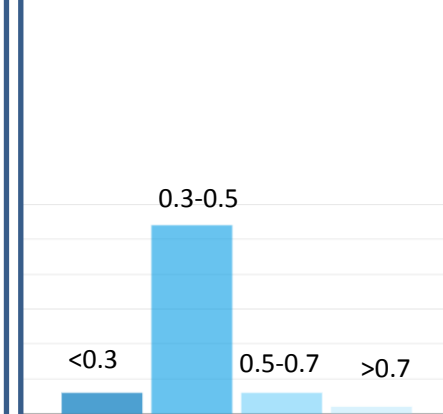
### Age at symptoms onset



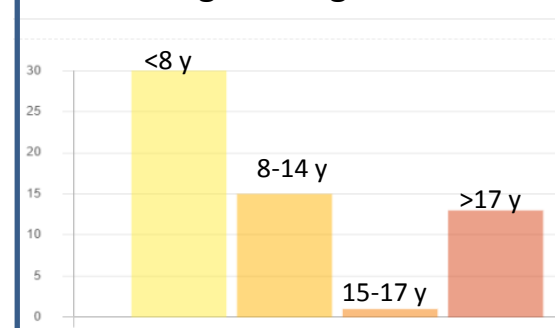
### Symptoms at onset



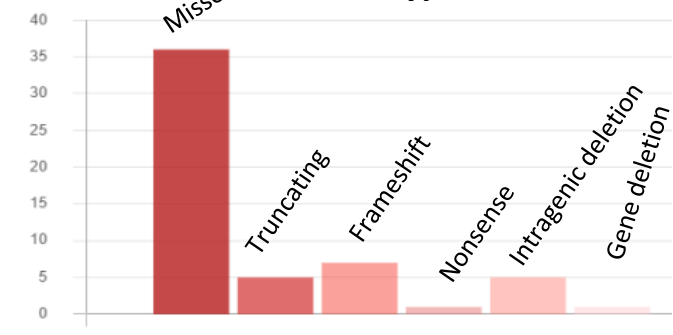
### CSF ratio



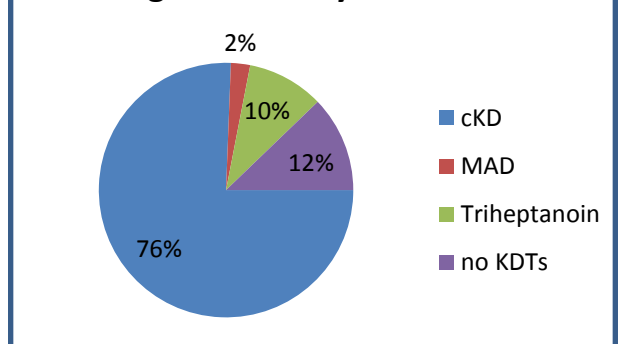
### Age at diagnosis



### Genotype



### Ketogenic Dietary Treatments



**Conclusions:** Collaboration between clinicians, researchers, advocacy groups, and patients can create essential community resource infrastructure to accelerate rare disease research. The Italian GLUT1DS Registry is an example of such an effort. Data collections will continue with the intent of raising awareness of the disease. It provides a unique resource for clinicians and researchers to identify unmet clinical needs, gaps in management and to address new treatment trials.