GenIDA – Genetic of Intellectual Deficiency Disorder and Autism: collecting and rendering the expertise of parents / carers

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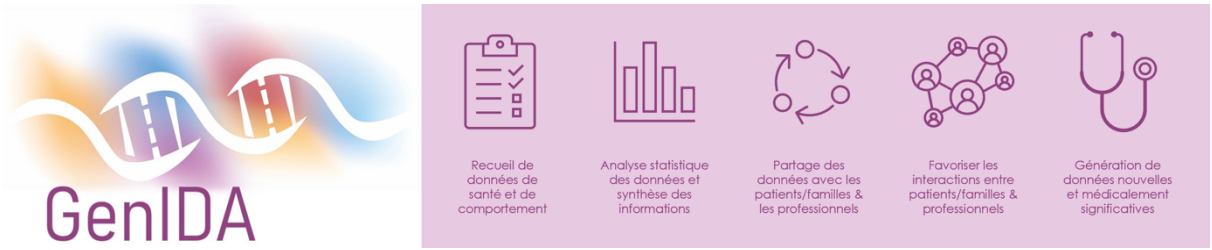

The GenIDA project is an international participatory research project on forms of intellectual deficiency, autism and genetic epilepsy, initiated by the Professor Mandel from the Strasbourg University, France.
This project aims to collect longitudinal health information from patient's families in France ad abroad, asking them to provide their observations via a structured questionnaire covering the various manifestations (medical, behavioral, etc.) of the disease in the patient and its natural history (evolution at different age). The clinical questionnaire is composed of 46 questions and is currently available online in 8 languages (English, Dutch, French, German, Italian, Portuguese, Greek and Spanish).
The unidentified collected data are available for volunteer professionals (clinicians, researchers, etc.) for analysis to create new knowledge significant medically which can translate to an improvement of care for concerned people. Results are also available summarized for non-profits and concerned Facebook groups, and for families having taken the questionnaire.

GenIDA includes almost 2000 participants, gathered in cohorts of various size, and almost 250 clinicians and reseachers registered as referral professional for the cohorts. The results of the analysis of aggregated data for specific cohorts, even small ones (n≥20 patients)[1], or cross-cohort[2], have been published in peer-reviewed journals. These results are consistent with the already published data in medical literature, while bringing a greater phenotypic richness in the pathologies studied[3], for instance regarding the sensitivity to certain co-morbidities or the perceived efficacy and/or adverse effects of specific treatments. We can cite the exemple of the Koolen-de-Vries (KdVS) syndrome for which the analysis of collected data from 237 patients showed aspects of the pathology unkown of professionals [4] leading to incidental studies to characterize musculoskeletal problems associate with KdVS[5] or aiming at broadening knowledge on its ophtalmological manifestations[6]. The medical data that emerged was transposed in the form of clinical management recommendations (*Professional Clinical*

[1] Durand et al. (2022) *Clinical Genetics*, https://doi.org/10.1111/cge.14190
[2] Coutelle et al. (2022) *BMC Psychiatry*, https://doi.org/10.1186/s12888-022-04213-6
[3] Ruault et al. (2024), *Molecular Genetics & Genomic Medicine*, https://doi.org/10.1002/mgg3.2363; Forbes*, Morison* et al. (2024), *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics* https://doi.org/10.1002/ajmg.b.32971
[4] Colin*, Burger* et al. (2023) *Genetics in Medicine Open*, https://doi.org/10.1016/j.gimo.2023.100817
[5] Bouman et al. (2023) *American Journal of Medical Genetics, A*, https://doi.org/10.1002/ajmg.a.63334
[6] Shalev et al. (2023) *Canadian Journal of Ophthalmology*, https://doi.org/10.1016/j.jcjo.2023.11.021

Practice Guidelines)[7] for the direct benefit of those most concerned, i.e. patients and their families.

[7] Thevenon, Marey, Gauthier, et al. (2023) PNDS - Syndrome de Koolen de Vries