# THE GenIDA NEWSLETTER

Number 1 / January 2024

## PUBLICATIONS 2023

Colin*, Burger*, et al. (2023) GenIDA, an international participatory study of medical and natural history data in genetic forms of neurodevelopmental disorders: novel observations in a large cohort of patients with Koolen-de Vries syndrome. Genetics in Medicine Open, https://doi.org/10.1016/j.gimo.2023.100817

Burger, et al. (2023) GenIDA: An international participatory database to gain knowledge on health issues related to genetic forms of neurodevelopmental disorders. Journal of Neural Transmission, https://doi.org/10.1007/s00702-022-02569-3

Mollereau, et al. (2023) A burning question from the first international BPAN symposium: is restoration of autophagy a promising therapeutic strategy for BPAN? Autophagy, https://doi.org/10.1080/15548627.2023.2247314

Bouman, et al. (2023) Clinical and radiological assessment of scoliosis in Koolen-de Vries syndrome, American Journal of Medical Genetics, A, https://doi.org/10.1002/ajmg.a.63334

Shalev, et al. (2023) Ocular Manifestations in Koolen-de Vries Syndrome – an International Study, Canadian Journal of Ophthalmology, https://doi.org/10.1016/j.jcjo.2023.11.021

Most of these articles are open access, and you just need to follow the link to download them; for the others, please contact us.

## EDITORIAL

First of all, let us wish you all the best for the New Year.

A new year means new projects, and to get things off to a good start, we thought we'd bring you the GenIDA newsletter to keep you up to date with our progress.

This first issue is essentially a review of our actions in 2023, a very fertile year, particularly in terms of publications concerning the data collected (see banner opposite). Also of note is the publication of the French Guidelines - Syndrome de Koolen-de Vries to which GenIDA contributed (Official Guidelines for the Diagnosis and Care of a Disease).

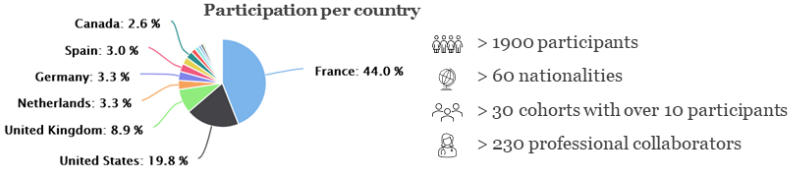
Many thanks to all of you, patients and families, for your involvement in GenIDA! Thank you for responding favourably to our requests, whether it's to answer a new questionnaire on the site, update your data, etc. We would also like to thank all the professionals who work with us to analyse the data we collect, thereby contributing to the development and smooth running of GenIDA.

The GenIDA team

*Many thanks to all of you, patients and families, for your involvement in GenIDA!*

## GenIDA IN KEY FIGURES

2023 was an excellent year for recruitment, with the threshold of 1,900 participating families exceeded last December, and more than 200 professionals showing an interest in analysing the data collected.



## 6th MEETING OF OUR INTERNATIONAL SCIENTIFIC ADVISORY BOARD

2023 was the perfect opportunity for GenIDA to resume the annual face-to-face meeting of our international Scientific Advisory Board at the European Society of Human Genetics conference, which took place from 10 to 13 June in Glasgow, Scotland. After opening the meeting with an update on the development of GenIDA since the last Scientific Advisory Board meeting (recruitment of participants, publications, etc.), our collaborators were able to talk about their experience in analysing the data collected via our database. Next on the agenda was a crucial point concerning improvements to the ergonomics of the GenIDA website, which would enable us to make further progress in recruiting participating families, as well as professionals involved in data analysis.

From left to right: Michael Patton, Londres, UK (représentant Patients Syndrome de Noonan); David Koolen, Nijmegen, NL ; Thomas Smol, Lille, FR ; Manon Chrétien, Strasbourg, FR ; Julien Thevenon, Grenoble, FR ; Klea Vyshka, Paris, FR ; Ece Gepni, Istanbul, TR ; Hulya Kayserili, Istanbul, TR ; Pauline Burger, Strasbourg, FR ; Tijtske Kleefstra, Rotterdam, NL ; Delphine Héron, Paris, FR ; Sylvie Odent, Rennes, FR ; Benjamin Durand, Strasbourg, FR ; Jean-Louis Mandel, Strasbourg, FR.

## 2024 PROJECTS

The major project for the new year will be the implementation of GenIDA's new web interface, which will be more ergonomic and user-friendly. The first mock-ups of the new interface were delivered to us in December, and a version of the site should be available by the summer of 2024.

Specific studies on the DDX3X, SLC6A1, POGZ, KBGS and SETD5 cohorts are currently underway; we will share the results as soon as possible (if you are concerned and interested, contact us to find out more).

## DON'T FORGET …

- To spread the word about GenIDA to other families and professionals, our flyer is available on request in electronic format (French, English, German, Dutch, Italian, Portuguese, Greek, Bulgarian, Romanian and Spanish) or can be downloaded directly from our website.
- To encourage other families to take part in the project if you can..
- To update your answers to the health questionnaire by logging back on to our website.

## CONTACT

GenIDA - IGBMC
1, rue Laurent Fries, 67400 Illkirch, France

Email : genida@igbmc.fr

Genida International Project

@GenIDAproject

Genida project

https://genida.unistra.fr/