IGPrare project

IGPrare project is a collaborative study founded on a partnership between an academic research team from Aix-Marseille University (“ADES” UMR 7268) and EURORDIS, and financed by the French agency of biomedicine. We work together with patient associations, geneticists, healthcare professionals, legal experts and researchers in human and social sciences.

Context

Patients with rare genetic diseases often have to get through a long medical Odyssey before being diagnosed. The diagnosis announcement is a major milestone in their lives, and it can also impact their relatives if the disease happens to be hereditary. In that case, at-risk relatives aware of their condition might benefit from preventive or curative measures. As a result, patients are often held responsible for ensuring accurate genetic information disclosure to their family, although being fragilized by the recent announcement. In practice, they might face multiple difficulties, as it involves a good understanding of the disease and its transmission mechanisms, as well as accurate communication skills. Not to mention that sometimes, patients also have to face their family rejection or denial, and even legal actions.

Objectives

1

To understand the circumstances and factors that can lead to a misunderstanding or a rejection of genetic information by relatives.

2

To propose accurate solutions for patients or professionals to improve the efficiency and acceptability of genetic information disclosure.

Conduct of the study

National scale

- **Understand** the mechanisms of genetic information disclosure by collecting French patients' detailed experiences through a questionnaire designed by patients associations, health professionals and researchers in human and social sciences.

European scale

- **Observe** the diversity of genetic information disclosure procedure across Europe *(including legal framework, healthcare system, culture...)* for a dozen of rare genetic diseases.
- **Share** the most efficient and acceptable solutions elaborated in different countries to bring new approaches and possible ways to improve the genetic information disclosure.

Step 1

- Interview Europeans patients associations representatives about the global patients' experiences of genetic information disclosure in their country.

- Disseminate a questionnaire about the legal framework and recommendations regarding genetic information disclosure to family, to healthcare professionals from each country involved in this study.

Step 2

Data statistical analyse.

Step 3

Discussion of both French and European findings to elaborate and share new solutions for patients and health professionals.

As our partner, we will keep you aware of the progress achieved and the study results by regularly publishing synthesis on our website.

www.igprare.fr

Results dissemination

At the end of the project, we will share our findings to :

Professionals and patients

To provide useful and practical solutions for patients to ensure genetic information disclosure.

Scientific community

To allow further research about this subject and confirmation of our findings by other researchers.

How ?

Through publications on our website and networks involving patients and professionals associations.

Through articles publications in scientific revues and posters presentations at national and European congresses.

Join us to make this project a success !

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IGP rare

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www.eurordis.org