The *IGPrare* European study : information disclosure to family in case of a rare genetic disease

M.Gottrau [1], A.Altavilla [2], S.Courbier [3], F.Faurisson[4], R.Favresse[3], A.Helm[3], F.Houyez[3], M.Mathieu [1,4]

***1**: ADES UMR7268, Aix-Marseille Université* ***2**: ERER-PACA Corse, Marseille* ***3**: Eurordis* ***4**: Tous Chercheurs*

Context

Patients diagnosed with a **rare genetic disease** are often considered a **moral duty** to inform their **at-risk family members**, in case they might benefit from treatments or prevention measures too.
However, disclosing such a personal information raises **ethical, legal and medical issues.** To answer them, countries adopted different approaches in order to protect **patients' right to confidentiality** and at-risk **family members' right to be informed** on their health.

Objectives :

- Understand the difficulties faced by patients to inform their relatives
- Review the diversity of solutions elaborated in Europe to frame genetic information disclosure to family in case of a rare disease.

Method

Dissemination of **2 surveys** in Europe :

Survey to **patients' associations' representatives** for different rare genetic diseases :

- Investigate the **real life conditions** of genetic information disclosure reported by patients.
- Filled during **interviews** by visioconference.

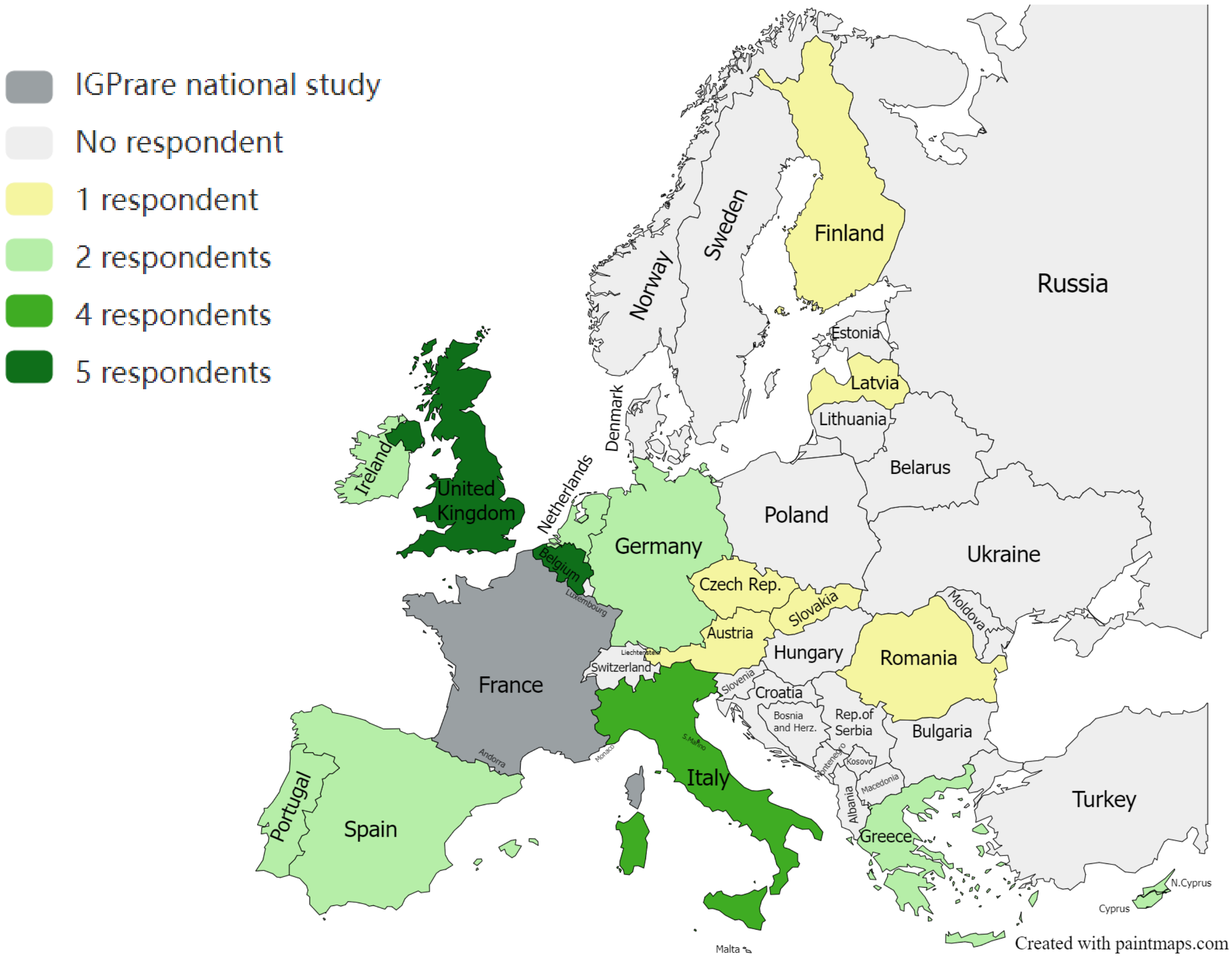
Survey to **healthcare professionals** in genetics :

- Investigate the **legal framework and ethical guidelines** in different countries.
- Filled online by participants.

Patients' associations' representatives survey

- **34 respondents in 16 countries.**
- **10 different diseases :**
  - 22q11 mutations *(6 associations)*
  - Cystic fibrosis *(6)*
  - Neuromuscular dystrophy *(5)*
  - Haemophilia *(4)*
  - Huntington *(4)*
  - Charcot-Marie-Tooth *(3)*
  - Friedrich ataxia *(2)*
  - Sickle cell anaemia *(2)*
  - Thalassemia *(2)*
  - X fragile *(1)*

Respondents to the associations survey

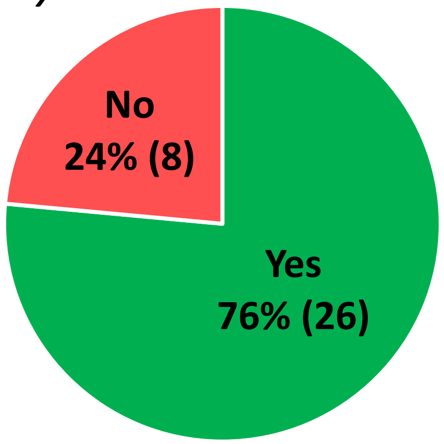


Patients' associations' representatives survey

The procedure of genetic information disclosure to family seems to be discussed in Europe *(Fig.* ***A****).*

A

Is genetic information disclosure to family a subject of discussions in your association? in your country?

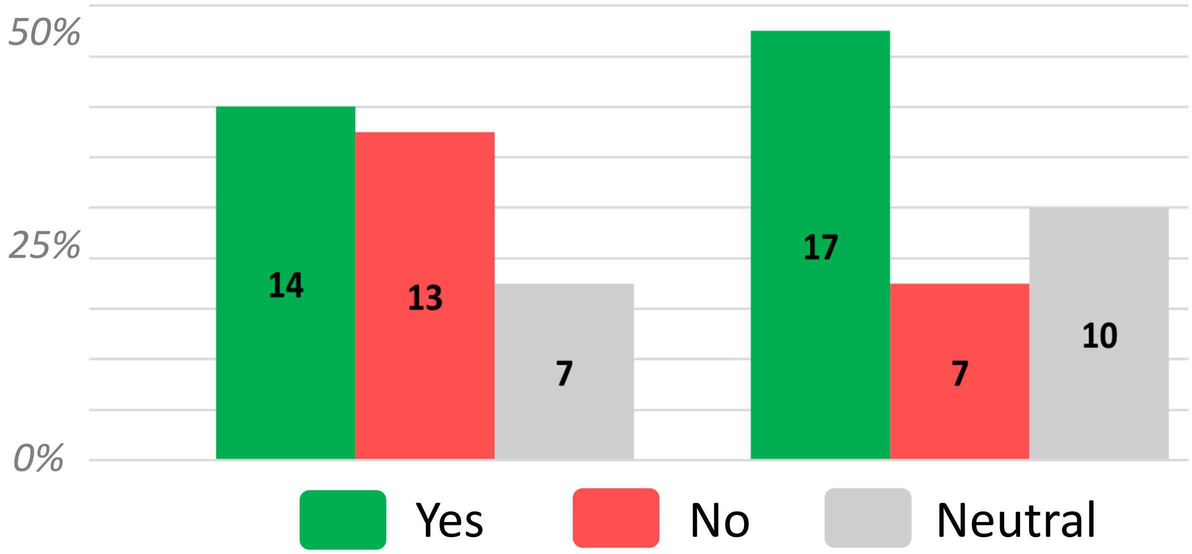


Regarding the current way to disclose genetic information, there are different representations of its efficiency to inform relatives and acceptability for patients and family *(Fig.* ***B****)*, even between people from a same country or for a same disease.

B

Is the current way to inform relatives efficient?

Acceptable?



Patients’ associations’ representatives survey

Possible improvements most frequently reported by associations :

Quality of the information given to patients :
Easier to understand, information about the disease, its consequences, the need to inform relatives…

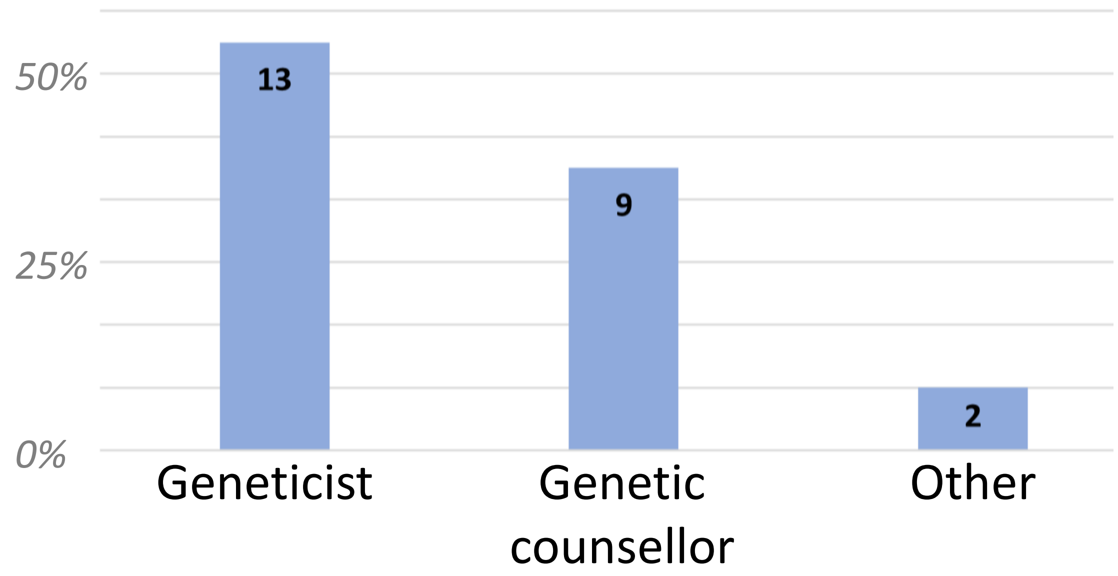
Quality of the healthcare professionals’ formation :
About rare diseases, empathy, dialogue, care relation…

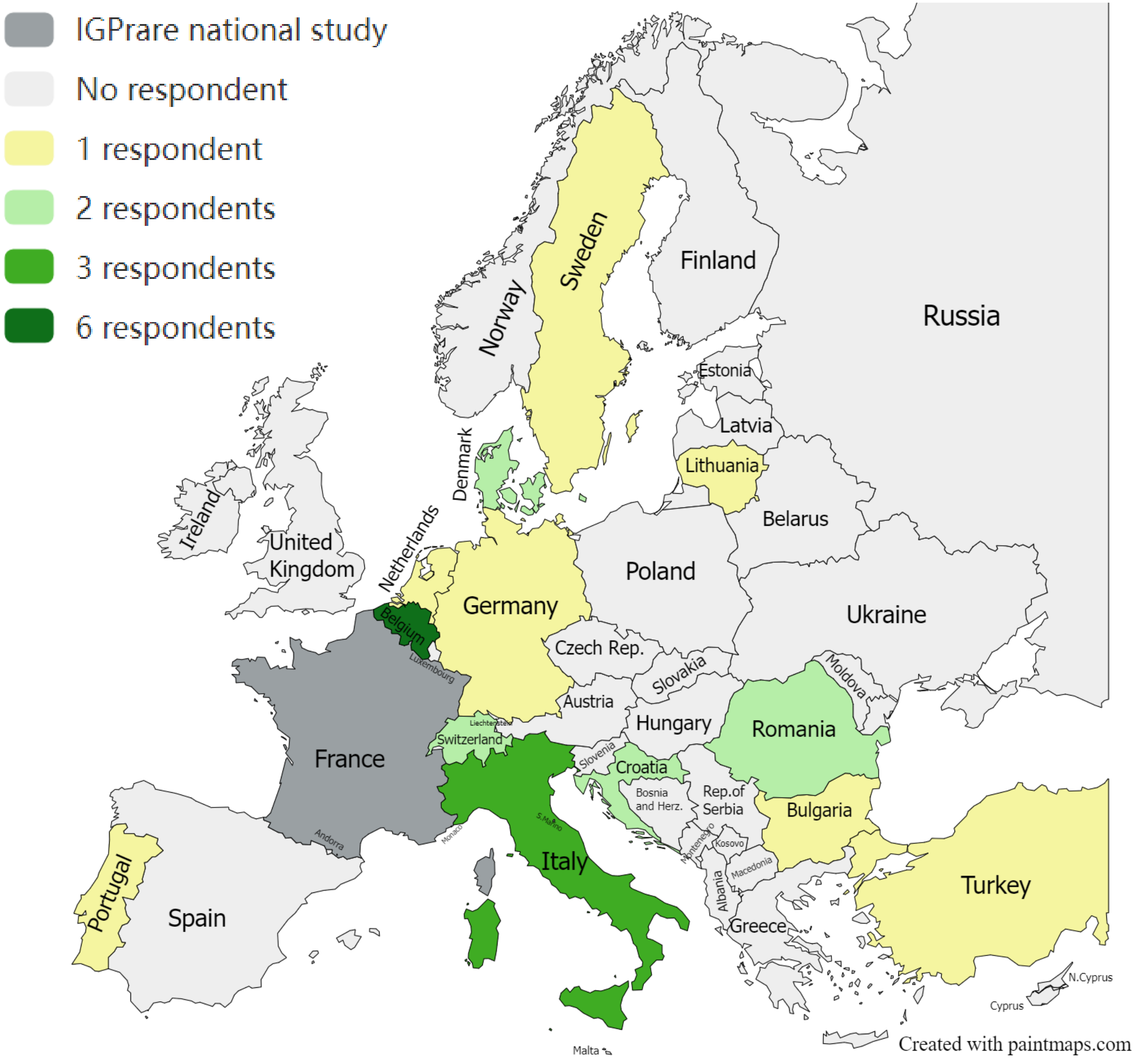
Information to public :
Public communication campaigns to inform about rare genetic diseases, to make it less taboo

Healthcare professionals survey

24 respondents in 13 countries.

C


Respondents' professions


Respondents to the healthcare professionals survey

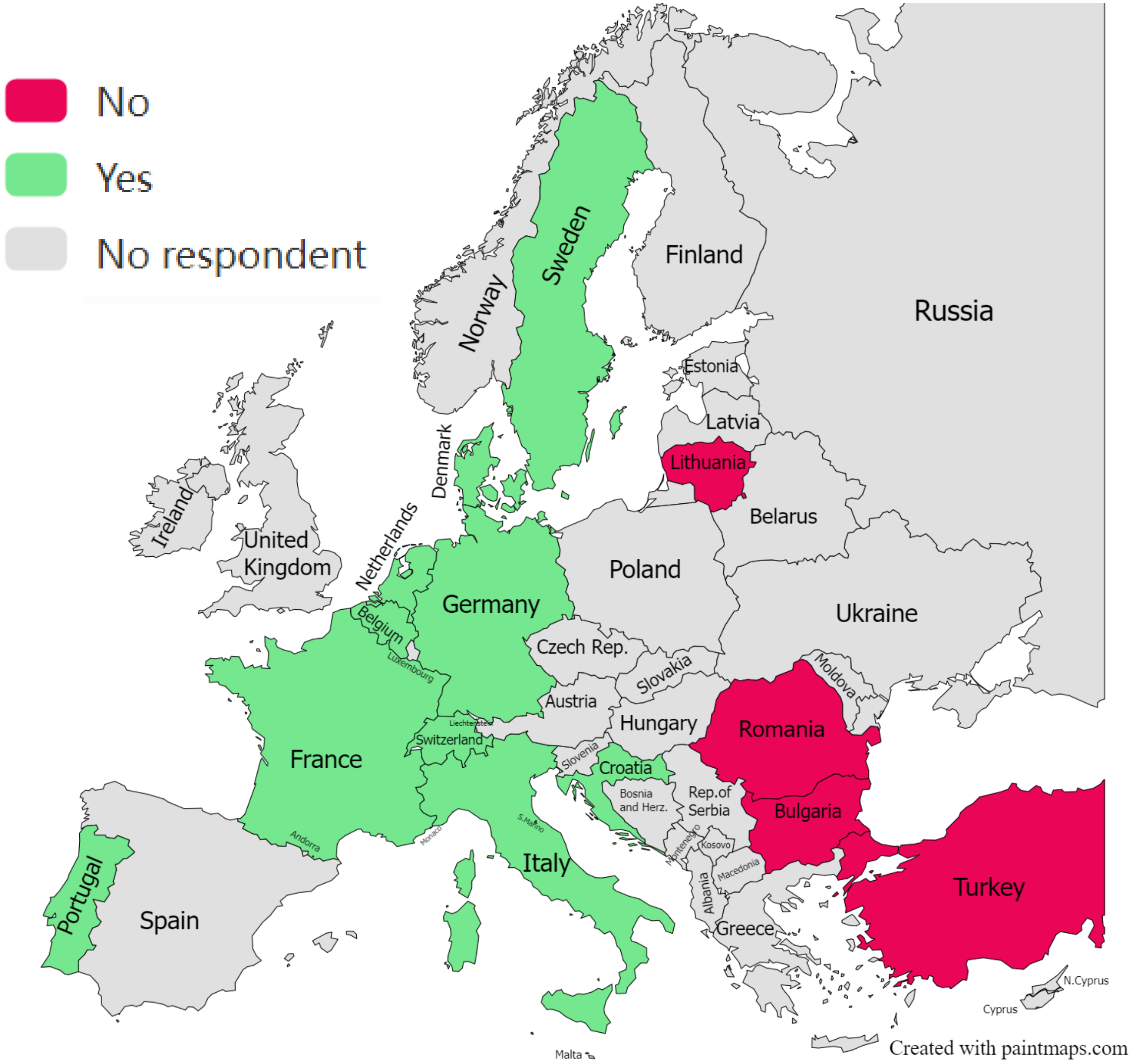
Created with paintmaps.com

Healthcare professionals survey

Existence of specific laws or guidelines to frame genetic information disclosure to family

In Western Europe countries, there seem to have more legislations or ethical guidelines to frame genetic information disclosure to family.

However, healthcare professionals from a same country might report different answers regarding the existence or content of such laws or guidelines.

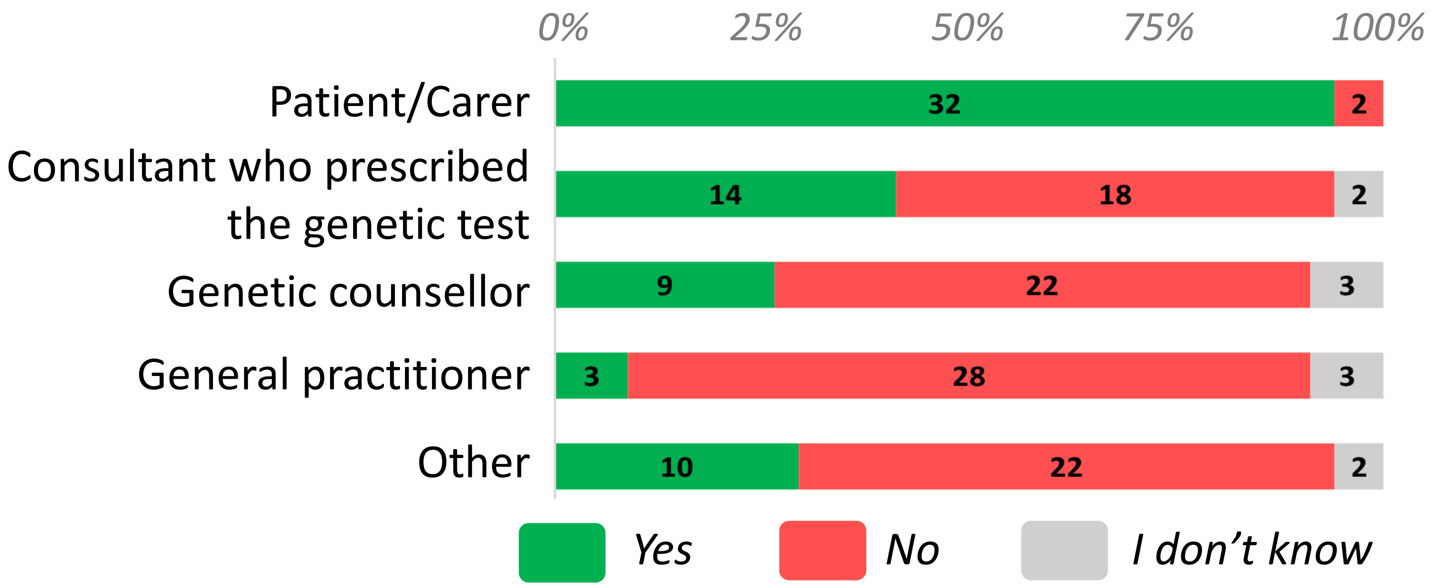


Who is responsible to inform relatives?

Regarding who is in charge of informing relatives, there are discrepancies between the legislation or guidelines reported by healthcare professionals, and the real life conditions of genetic information disclosure described by associations *(Fig. **D**).*

Patients' associations' representatives survey

D *Who is in charge of informing relatives?*



Healthcare professionals survey

- Differences according to countries' legal frameworks and guidelines.
- There are sometimes discrepancies between healthcare professionals from a same country.

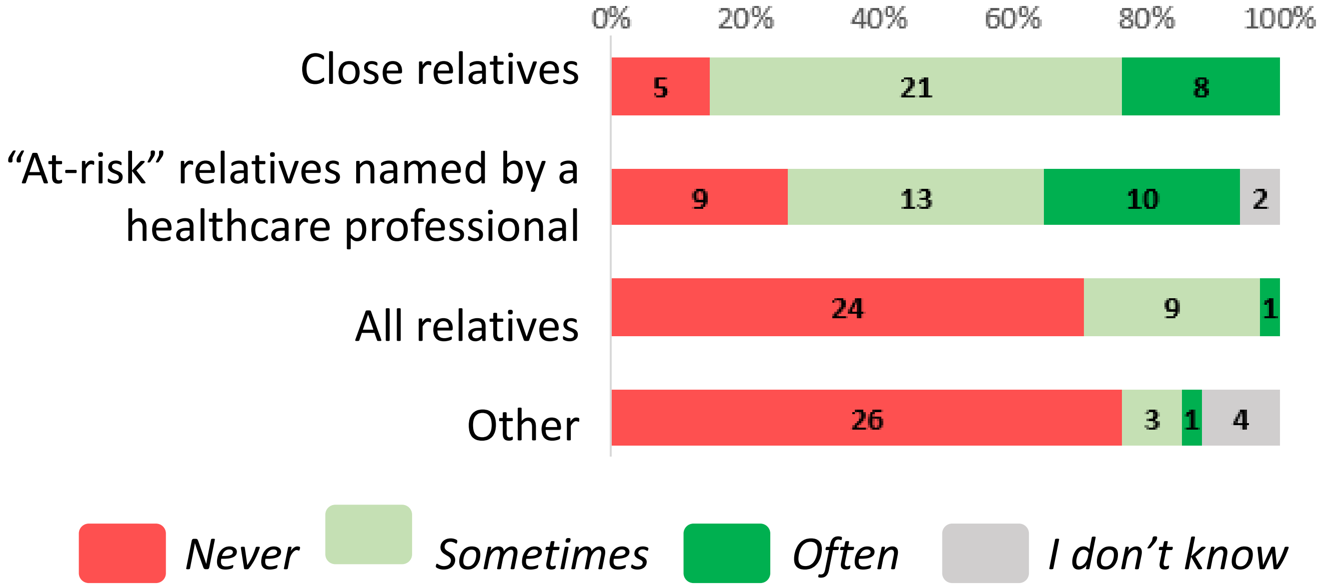
Which relatives are informed?

Genetic information seems to be disclosed in priority to close relatives *(Fig.* ***E****)*, rather than to "at-risk" relatives, like stated in the laws or guidelines *(Fig.* ***F****).*

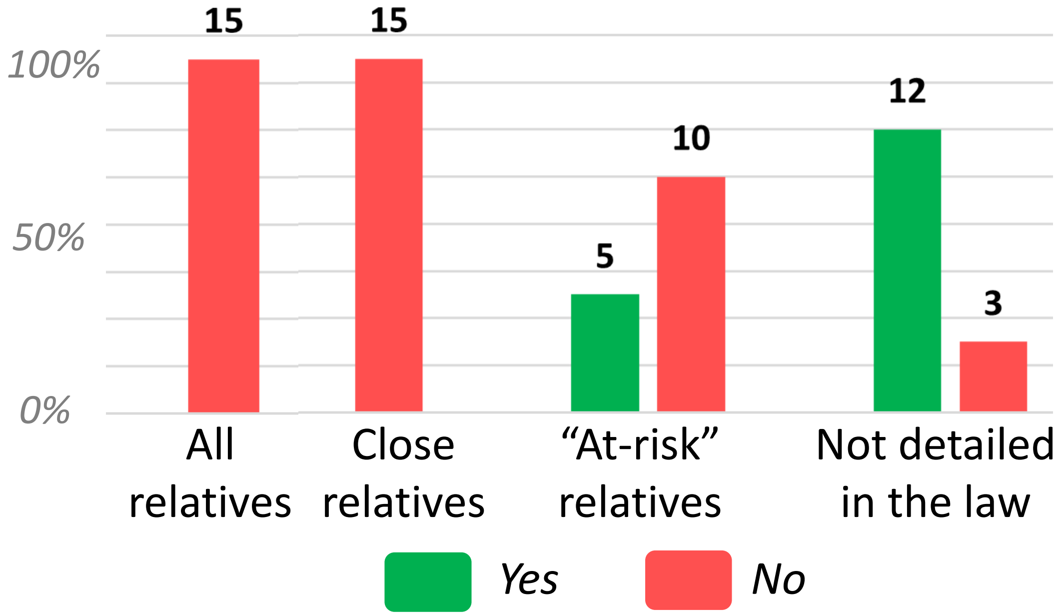
Patients' associations' representatives survey

Healthcare professionals survey

E *Which relatives are informed in general?*



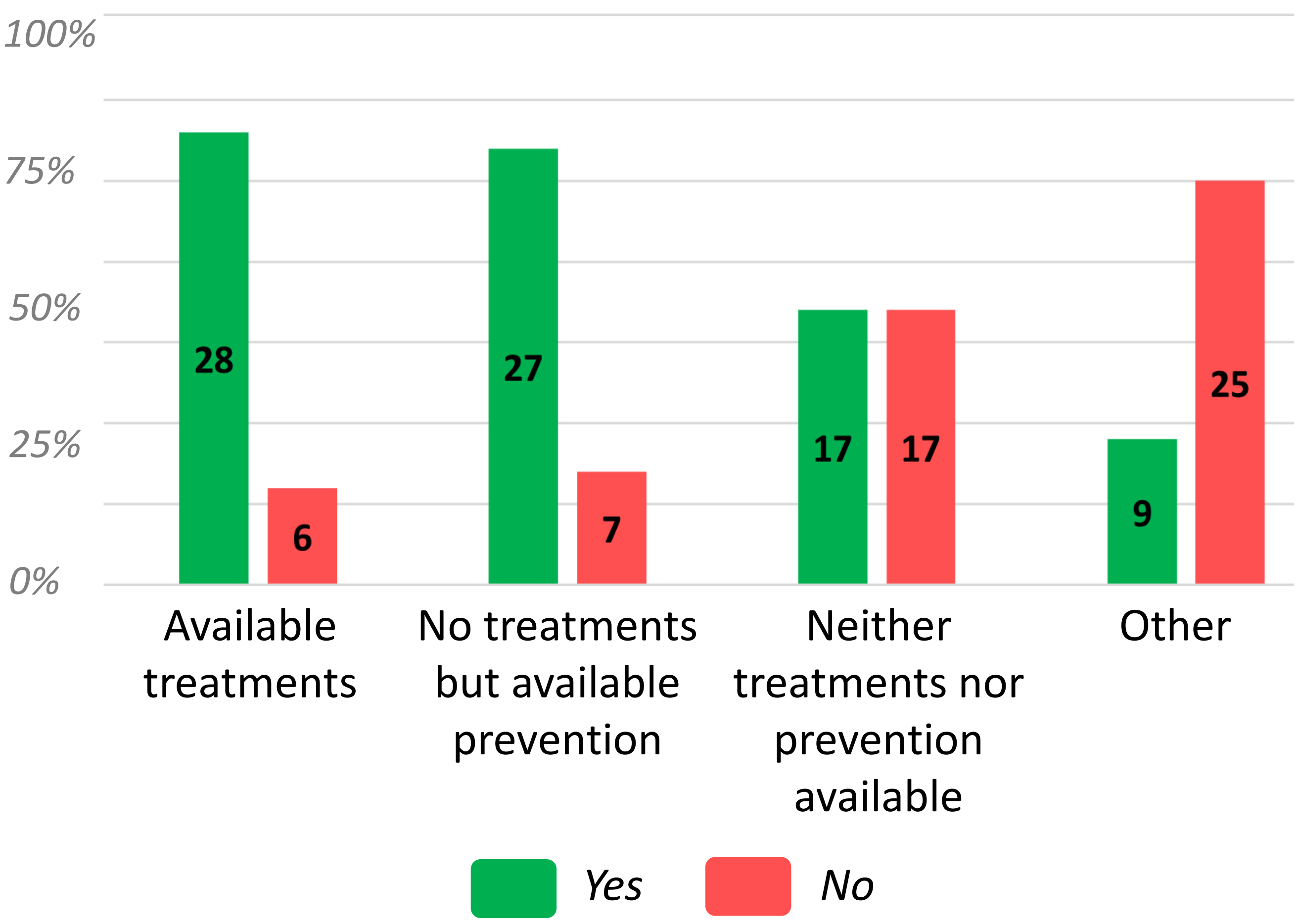
F *Which relatives should be informed according to the law?*



When is genetic information disclosed?

Patients' associations' representatives survey

- In case of available treatments or prevention, genetic information is disclosed most part of the time.
- If case there are not, relatives are informed half of the time.



G

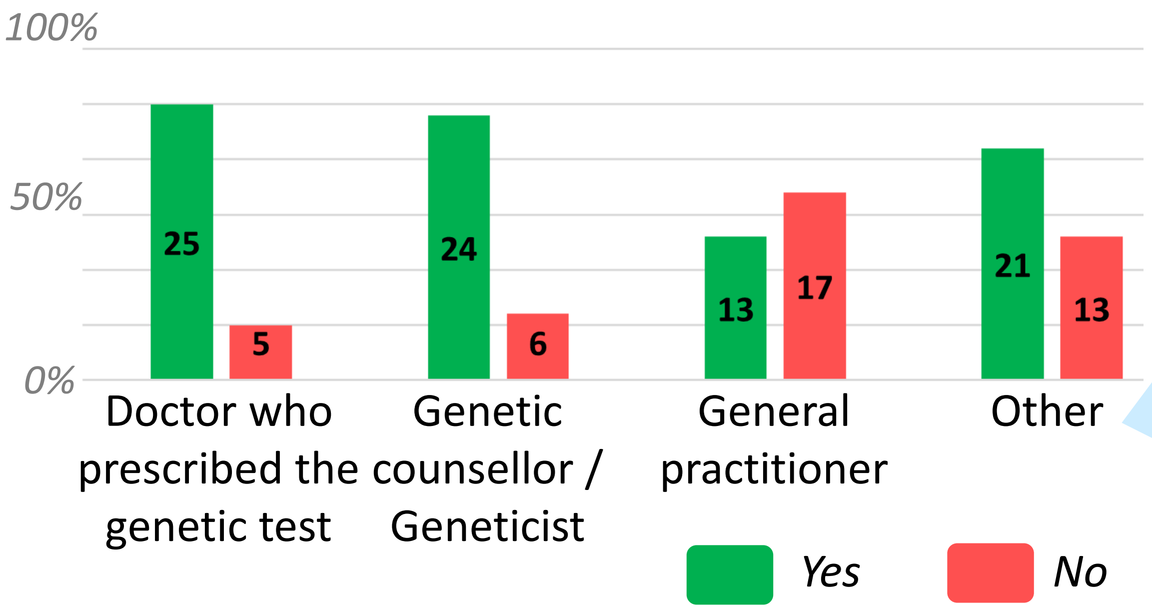
What situations justify to disclose genetic information to relatives?

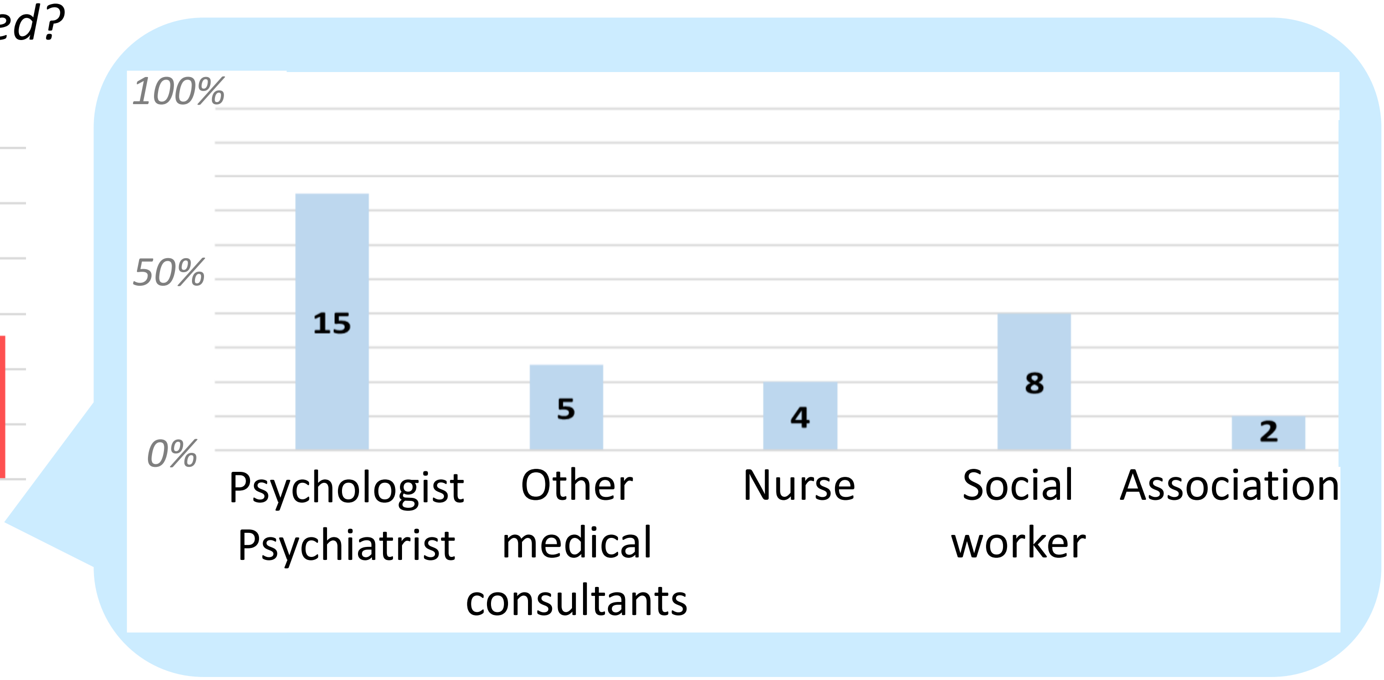
Expected healthcare professionals’ support

Almost all associations stated the importance of healthcare professionals’ support in order to disclose genetic information to relatives, especially psychological support.

Patients’ associations’ representatives survey

H *What type of professional should be involved?*





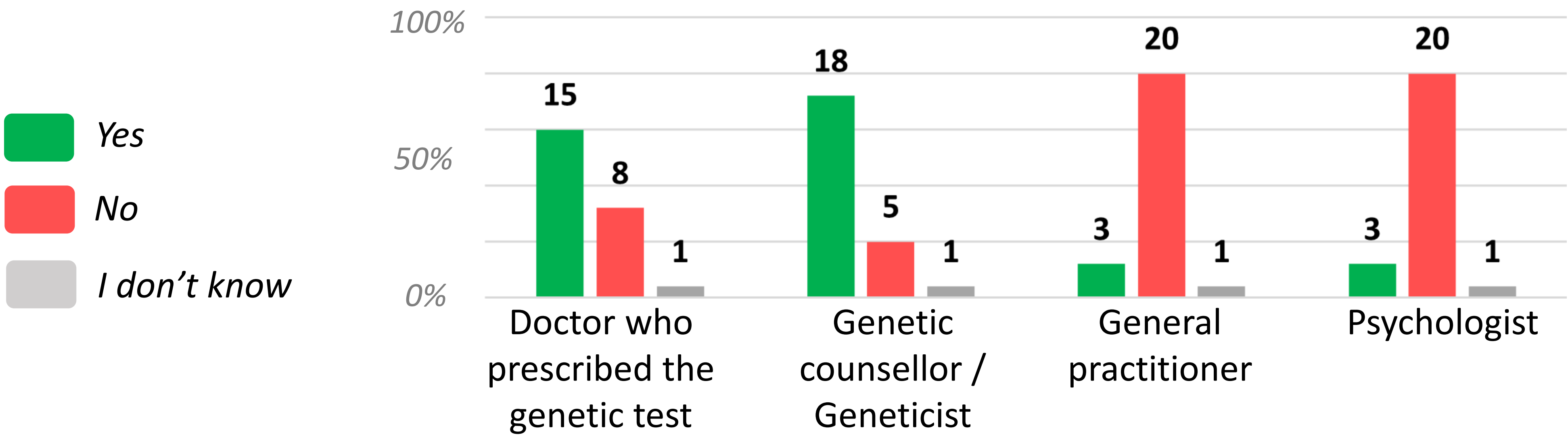
Real-life healthcare professionals’ support

However psychologists seems to be rarely involved according to the healthcare professionals *(Fig. **I**)*.

Healthcare professionals survey

I

What type of professional is involved in the procedure?



Conclusion

- This study enlightened the **diversity of patients' experiences** to disclose genetic information to their relatives in case of a rare disease. Patients from different countries seem to face **similar difficulties**, as well as **specific difficulties** due to a national or cultural context.

- There also seem to have **discrepancies between laws and guidelines** framing genetic information disclosure to family, and its **real-life conditions**.

These findings will be confronted to a national scaled study in France, involving more participants. The final objective is to **identify and share the most efficient and acceptable solutions** to improve practices in Europe, in order to **facilitate genetic information disclosure to family** in the future.

Contact : mariongott@gmail.com

Research study founded by the French Agency of Biomedicine ("Recherche 2020", convention 20AMP014) and approved by the Aix-Marseille-University's Ethics committee (decision n°2021-07-08-09).