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ORIGINAL ARTICLE

# Evaluation of the template letter regarding the disclosure of genetic information within the family in France

Cécile Zordan[1] · Laetitia Monteil[2] · Emmanuelle Haquet[3] · Christophe Cordier[4] · Eva Toussaint[1] · Pauline Roche[1] · Virginie Dorian[1] · Aline Maillard[5] · Edouard Lhomme[5] · Laura Richert[5] · Laurent Pasquier[6] · Linda Akloul[6] · Nicolas Taris[7] · Didier Lacombe[1,8]

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## Abstract

The 2011 French Bioethics Law regarding disclosure of genetic information within families enables health professionals to notify any at-risk relatives directly, with the patient's consent, using a template letter. To assess the impact of this template letter in terms of understanding, personal feelings and intent to contact a health professional, we conducted a study interviewing patients, members of the public and genetic professionals. Although the main response to the letter was anxiety, this was associated with good understanding of the content and most individuals mentioned intention to contact a health professional.

**Keywords** Genetic information disclosure · Template letter · Family communication · Responsibility · Genetic counselling · Ethical issues

## Introduction

The principles common to most guidelines about the communication of genetic information to families include the individual's moral obligation to communicate genetic information to their at-risk relatives, the health care professional's (HCP) encouragement to communicate this information to any at-risk relatives and the support given to the individual throughout this communication process (Forrest et al. 2007). In France, whenever a patient is diagnosed with a genetic anomaly, the HCP has the duty to inform the patient of risks to relatives. It remains the patient's decision whether or not to communicate this information to their relatives, but HCPs must inform the patient of the potential consequences for the health of their relatives if they do not disclose (Law no. 2004-800 of 6 August 2004). Communication among family members can be complex and result in conflicting senses of responsibility (Nycum et al. 2009), while the patient may feel ashamed, guilty or embarrassed about suffering from an inherited disorder (Parker and Lucassen 2003). The HCP must take into consideration family dynamics so as to improve the communication process (Dheensa et al. 2016), and given the family-related and ethical implications of a genetic diagnosis,

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✉ Cécile Zordan
cecile.zordan@chu-bordeaux.fr

1 Service de Génétique Médicale, Bâtiment Ecole de Sages-Femmes, Groupe Hospitalier Pellegrin, CHU de Bordeaux, Place Amélie Raba-Léon, 33 076 Bordeaux Cedex, France

2 Service de Génétique Médicale, CHU de Toulouse, Toulouse, France

3 Service de Génétique Médicale, CHRU de Montpellier, Montpellier, France

4 Département de Diagnostic Moléculaire, Synlab Genetics, Lausanne, Switzerland

5 Unité de Soutien Méthodologique à la Recherche Clinique et Epidémiologique, Service d'Information Médicale, CHU de Bordeaux, Bordeaux, France

6 Service de Génétique Clinique, CHU de Rennes, Rennes, France

7 Service de Génétique Oncologique, CRLCC Paul Strauss, Strasbourg, France

8 Inserm U1211, Université de Bordeaux, Bordeaux, France

incorporating counselling and education about family communication is a prerequisite for HCP practice (Forrest et al. 2010).

If the patient refuses to inform their at-risk relatives, the HCP is faced with a conflict between the respect of patient confidentiality, privacy and autonomy, and the potential harm prevention and health benefits for the relatives (Dheensa et al. 2016). The moral obligation to communicate medical information stands in opposition to the right to confidentiality. The international ethical guidelines recommend that HCPs should respect patient's confidentiality and should not contact at-risk relatives directly (Forrest et al. 2007). The American Society of Human Genetics Social Issues Subcommittee (1998) policy is that genetic health professionals can ethically breach confidentiality by disclosing the genetic information without the patient's consent, in exceptional cases, when "the harm is highly likely to occur and is serious, imminent and foreseeable; the at-risk relative(s) is identifiable; and the disease is preventable, treatable or medically accepted standards indicate that early monitoring will reduce the genetic risk". The ASHG statement about disclosure is an exceptional status based on the Tarasoff legal case (Cal. 1976). In the UK and Australia, there has been no specific legislation regulating disclosure by HCP without the patient's consent (Clarke et al. 2005). A recent review (Dheensa et al. 2016) described that patients and the public have an unfavourable view of HCPs sharing information without consent.

In France, the HCP is not legally allowed to communicate genetic information to relatives without the patient's consent. The Bioethics Law includes the "medical information procedure" (Law no. 2011-814 of 7 July 2011, art. 2): the patient must inform their at-risk relatives after a genetic diagnosis of a serious condition that allows preventive or care measures "and genetic counselling" (Legislative decree no. 2013-527). However, if the patient agrees to share genetic information with relatives, but does not wish to do this themselves, they may authorise the HCP to do so. In this case, the HCP sends a registered letter to at-risk relatives, without revealing the patient's identity or genetic anomaly. The decree (Legislative order of 20 June 2013) includes a specific template letter. This letter encourages relatives to make an appointment at a genetics centre and the template can be adapted by the HCP as they deem fit. If the patient does not respect the obligation to inform their relatives, they become liable, can be held responsible according to civil law and fined by a judge for the damage caused (Legislative order of 8 December 2014).

A previous study showed that HCPs do not use the same family information procedure for every disease and family situation (d'Audiffret and de Montgolfier 2016). The template letter in the annex on the decree informs relatives about the genetic risk and the means to contact a genetics centre. Some genetics centres have drafted their own template letter (Lahlou-Laforet et al. 2014), but to date, none of these templates have been evaluated. Overall, it is currently unclear whether the national template letter is well enough understood and accepted by genetics professionals and patients or whether an alternative template would ensure better understanding and acceptability.

### Aim of the study

The aim of this study was to assess the national template letter (Supplementary document), as well as an alternative template (Supplementary document), in terms of understanding, feelings and intent to contact a HCP among both patients consulting in genetics centres and the general population.

## Methods

We conducted a quantitative study that consisted of three surveys, each of which included some open-ended questions. The study was performed by six genetic counsellors practicing their profession in five departments of medical genetics and/or oncogenetics in France (Bordeaux, Montpellier, Rennes, Strasbourg and Toulouse). An approval for this study was granted by the ethical committee CPP SOOM III. Eligible participants were French-speaking and aged 18 and above. Oral informed consent was obtained from all individual participants included in the study. The participants were informed they could withdraw from the study whenever they wished. The patients knew they could refuse to participate without their medical care being affected. The study was divided into three phases.

For study 1 (assessment of the impact of decree template letter), two groups of participants were recruited between September and December 2013 by six genetic counsellors: patients who attended a genetic counselling consultation with one of these genetic counsellors and individuals from the general population.

A semi-structured questionnaire was created by these genetic counsellors, which focused on understanding, feelings and the intent to contact a HCP. Genetic counsellors administered the survey to individuals after they read the template letter contained in the decree of 20 June 2013 (letter A in the Supplementary document). This questionnaire included 19 questions: five questions about the understanding of letter, five questions about the feelings, two questions to assess the intent to contact a HCP and six questions about characteristics of participants (Table 1). The following data were recorded: gender, age, socio-professional category, number of children and work in the health care domain. The primary outcome measure was the understanding of letter A. It was evaluated by the answer of the fourth question with a binary variable (Yes/No): if the response c, d or e was ticked, we considered that the participant had well understood the meaning of the

**Table 1** Questionnaire of studies 1 and 2

| | | | Letter A | | | | | | Letter B | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | | | Patient | | General pop. | | Total | | Patient | | General pop. | | Total | |
| | | | *N* (75) | % | *N* (73) | % | *N* (148) | % | *N* (75) | % | *N* (80) | % | *N* (155) | % |
| Characteristics of participants | Sex | Female | 46 | 61.3 | 42 | 57.5 | 88 | 59.5 | 48 | 64.0 | 52 | 65.0 | 100 | 64.5 |
| | | Male | 29 | 38.7 | 31 | 42.5 | 60 | 40.5 | 27 | 36.0 | 28 | 35.0 | 55 | 35.5 |
| | Age (median) | | 35 | | 37 | | 35.5 | | 41 | | 40 | | 40 | |
| | Had at least one child | | 51 | 68.0 | 46 | 63.0 | 97 | 65.5 | 49 | 65.3 | 48 | 60.0 | 97 | 62.6 |
| | Worked in the health care sector | | 13 | 17.3 | 26 | 35.6 | 39 | 26.4 | 12 | 16.0 | 20 | 25.0 | 32 | 20.6 |
| Understanding | 1. How many times did you read this letter? | | | | | | | | | | | | | |
| | One time | | 58 | 77.3 | 66 | 90.4 | 124 | 83.8 | 54 | 72.0 | 82 | 77.5 | 116 | 74.8 |
| | Two times | | 17 | 22.7 | 7 | 9.6 | 24 | 16.2 | 19 | 25.3 | 17 | 21.3 | 36 | 23.1 |
| | Three times or more | | 0 | 0.0 | 0 | 0.0 | 0 | 0.0 | 2 | 2.7 | 1 | 1.3 | 3 | 1.9 |
| | 2. What is your level of understanding of this letter? | | | | | | | | | | | | | |
| | 0. Nothing is understood | | 0 | 0.0 | 0 | 0.0 | 0 | 0.0 | 1 | 1.3 | 1 | 1.3 | 2 | 1.3 |
| | 1 | | 0 | 0.0 | 0 | 0.0 | 0 | 0.0 | 0 | 0.0 | 0 | 0.0 | 0 | 0.0 |
| | 2 | | 3 | 4.0 | 7 | 9.6 | 10 | 6.8 | 0 | 0.0 | 0 | 0.0 | 0 | 0.0 |
| | 3 | | 13 | 17.3 | 1 | 1.4 | 14 | 9.5 | 3 | 4.0 | 4 | 5.0 | 7 | 4.5 |
| | 4 | | 9 | 12.0 | 19 | 26.0 | 28 | 18.9 | 16 | 21.3 | 11 | 13.8 | 27 | 17.4 |
| | 5. All are understood | | 50 | 66.7 | 46 | 63.0 | 96 | 64.9 | 55 | 73.3 | 64 | 80.0 | 119 | 76.8 |
| | 3. Are there paragraphs which you had difficulty understanding? | | | | | | | | | | | | | |
| | a. No | | 51 | 68.0 | 57 | 78.1 | 108 | 73.0 | 66 | 88.0 | 71 | 88.8 | 137 | 88.4 |
| | b. Yes | | 24 | 32.0 | 16 | 21.9 | 40 | 27.0 | 9 | 12.0 | 9 | 11.3 | 18 | 11.6 |
| | 4. What do you *mainly* learn through this letter? | | | | | | | | | | | | | |
| | a. Nothing | | 1 | 1.3 | 0 | 0.0 | 1 | 0.7 | 2 | 2.7 | 0 | 0.0 | 2 | 1.3 |
| | b. I am a carrier of a genetic anomaly | | 2 | 2.7 | 1 | 1.4 | 3 | 2.0 | 5 | 6.7 | 0 | 0.0 | 5 | 3.2 |
| | c. A member of my family is a carrier of a genetic anomaly | | 31 | 41.3 | 33 | 45.2 | 64 | 43.2 | 28 | 37.5 | 29 | 36.3 | 57 | 36.8 |
| | d: I could be a genetic disease carrier | | 25 | 33.3 | 23 | 31.5 | 48 | 32.4 | 27 | 36.0 | 37 | 46.3 | 64 | 41.3 |
| | e. A consultation with a geneticist is offered to me | | 16 | 21.3 | 15 | 20.5 | 31 | 20.9 | 13 | 17.3 | 13 | 16.3 | 26 | 16.8 |
| | f. It is an unsolicited letter | | 0 | 0.0 | 1 | 1.4 | 1 | 0.7 | 0 | 0.0 | 1 | 1.3 | 1 | 0.6 |
| | 5. Do you think that all paragraphs are useful? | | | | | | | | | | | | | |
| | a. Yes | | 63 | 84.0 | 54 | 74.0 | 117 | 79.1 | 70 | 93.3 | 74 | 94.9 | 144 | 94.1 |
| | b. No | | 12 | 16.0 | 19 | 26.0 | 31 | 20.9 | 5 | 6.7 | 4 | 5.1 | 9 | 5.9 |
| Feelings | 6. The reading of this letter may arouse different feelings. What is the feeling that best applies to you? | | | | | | | | | | | | | |
| | a. Happy | | 0 | 0.0 | 1 | 1.4 | 1 | 7.0 | 0 | 0.0 | 0 | 0.0 | 0 | 0.0 |
| | b. Serene | | 2 | 2.7 | 8 | 11.0 | 10 | 68.0 | 4 | 5.3 | 2 | 2.5 | 6 | 3.9 |
| | c. Curious | | 15 | 20.0 | 15 | 20.5 | 30 | 20.3 | 20 | 26.7 | 15 | 18.8 | 35 | 22.6 |
| | d. Emotionless | | 5 | 6.7 | 0 | 0.0 | 5 | 34.0 | 0 | 0.0 | 4 | 5.0 | 4 | 2.6 |
| | e. Shocked | | 4 | 5.3 | 5 | 6.8 | 9 | 6.1 | 3 | 4.0 | 3 | 3.8 | 6 | 3.9 |
| | f. Worried | | 45 | 60.0 | 35 | 47.9 | 80 | 54.1 | 41 | 24.7 | 44 | 55.0 | 85 | 54.8 |
| | g. Very worried | | 3 | 4.0 | 9 | 12.3 | 12 | 8.1 | 6 | 8.0 | 12 | 15.0 | 18 | 11.6 |
| | h. Angry | | 1 | 1.3 | 0 | 0.0 | 1 | 0.7 | 1 | 1.3 | 0 | 0.0 | 1 | 0.6 |
| | 7. If you feel worried, is it ….? (multiple answers can be selected) | | *N* = 48 | | *N* = 44 | | *N* = 92 | | *N* = 47 | | *N* = 56 | | *N* = 103 | |
| | a. For yourself | | 22 | 45.8 | 28 | 63.6 | 50 | 54.3 | 29 | 61.7 | 44 | 78.6 | 73 | 78.9 |

**Table 1** (continued)

| | | Letter A | | | | | | Letter B | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | | Patient | | General pop. | | Total | | Patient | | General pop. | | Total | |
| | | *N* (75) | % | *N* (73) | % | *N* (148) | % | *N* (75) | % | *N* (80) | % | *N* (155) | % |
| | b. For your child | | | | | 32 | 34.8 | | | | | 45 | 43.7 |
| | c. For your husband/wife | 2 | 4.2 | 1 | 2.3 | 3 | 3.3 | 5 | 10.6 | 3 | 5.4 | 8 | 7.8 |
| | d. For the affected family member | 22 | 45.8 | 16 | 36.4 | 38 | 41.3 | 27 | 57.4 | 27 | 48.2 | 54 | 52.4 |
| | 8. If you feel angry, is it …? | | | | | | | | | | | | |
| | a. In general | 0 | | 0 | | 0 | | 1 | | 0 | | 1 | |
| | b. Against affected family member | 1 | | 0 | | 1 | | 0 | | 0 | | 0 | |
| | c. Against the physician who sent the letter | 0 | | 0 | | 0 | | 0 | | 0 | | 0 | |
| | 9. What do you think about the term "genetic anomaly"? | | | | | | | | | | | | |
| | a. It is appropriate | 39 | 52.0 | 31 | 42.5 | 70 | 47.3 | 34 | 45.3 | 36 | 45.0 | 70 | 45.2 |
| | b. It is too frightening | 1 | 1.3 | 3 | 4.1 | 4 | 2.7 | 2 | 2.7 | 8 | 10.0 | 10 | 6.5 |
| | c. It is frightening but necessary | 24 | 32.0 | 31 | 42.5 | 55 | 37.2 | 30 | 40.0 | 26 | 32.5 | 56 | 36.1 |
| | d. It is reassuring | 1 | 1.3 | 1 | 4.1 | 2 | 1.4 | 0 | 0.0 | 2 | 2.5 | 2 | 1.3 |
| | e. Other | 10 | 13.3 | 7 | 9.6 | 17 | 11.5 | 9 | 12.0 | 8 | 10.0 | 17 | 11.0 |
| | 10. What do you think of the sentence "Some members may wish to discuss this and others will prefer to remain silent"? | | | | | | | | | | | | |
| | a. It is appropriate | 46 | 61.3 | 32 | 43.8 | 78 | 52.7 | | | | | | |
| | b. It is comforting | 4 | 5.3 | 5 | 6.8 | 9 | 6.1 | | | | | | |
| | c. It is unnecessary | 19 | 25.3 | 27 | 37.0 | 46 | 31.1 | | | | | | |
| | d. Other | 6 | 8.0 | 9 | 12.3 | 15 | 10.1 | | | | | | |
| Intention to contact HCP | 11. Will you contact a health care professional? | | | | | | | | | | | | |
| | a. Yes | 69 | 92.0 | 71 | 97.0 | 140 | 94.6 | 67 | 89.3 | 67 | 83.8 | 134 | 86.5 |
| | b. No | 3 | 4.0 | 2 | 2.7 | 5 | 3.4 | 2 | 2.7 | 2 | 2.5 | 4 | 2.6 |
| | c. Do not know yet | 3 | 4.0 | 0 | 0.0 | 3 | 2.0 | 6 | 8.0 | 11 | 13.8 | 17 | 11.0 |
| | 12. If yes, would you contact …? | | | | | | | | | | | | |
| | a. Your general practitioner | 22 | 29.3 | 9 | 12.3 | 31 | 20.9 | 7 | 9.3 | 13 | 16.3 | 20 | 12.9 |
| | b. A genetics department | 30 | 40.0 | 44 | 60.3 | 74 | 50.0 | 25 | 33.3 | 34 | 42.5 | 59 | 38.1 |
| | c. Your general practitioner and genetics department | 17 | 22.6 | 16 | 21.9 | 33 | 22.3 | 34 | 45.3 | 18 | 22.5 | 52 | 33.5 |
| | d. Another health professional | 0 | 0.0 | 2 | 2.7 | 2 | 1.4 | 1 | 1.3 | 2 | 2.5 | 3 | 1.9 |

letter. If not, we considered that the participant did not understand the letter. The secondary outcomes were the feelings after reading letter A and the recognition that they should contact a HCP if they were to receive such a letter personally. The feelings of the participants after reading the letter were evaluated by the answer of the sixth, seventh and eighth questions. The intention to contact HCP was evaluated by the answer of the 11th and 12th questions. A proportion higher or equal to 80% of understanding of the letter by the participants was expected and defined as clinically relevant. With a sample size of 75 participants, the estimation of the expected proportion of 80% (60/75) will have an adequate precision with 95% CI = (69%; 88%).

Based on these results, we recruited six genetic counsellors, one medical geneticist, two psychologists, one general practitioner, and five representatives of patient associations (*Vaincre les Maladies Lysosomales*, *Association Française du Syndrome de Rubinstein-Taybi*, *Association Française Ataxie de Freidreich*, *Connaître les Syndromes Cérébelleux*, *Alliance Maladies Rares*) to a discussion in order to design a new template letter (letter B in the Supplementary document). It was decided that paragraph 5 (P5) should be removed from the original template, that the appropriate law to this procedure should be cited and that the letter should be shortened.

In the second phase of the study, we evaluated letter B with the same methodology and the same questionnaire (without the tenth question) as letter A in study 1 (Table 1). The participants (patients who attended a genetic counselling consultation and individuals from the general population) were recruited between July and December 2015 by the genetic counsellors. The primary and secondary outcome measures were the same as in study 1.

For study 3 (comparison of the two letters), an online survey was created with a randomised reading order of the two letters (A and B). We recruited participants who were either patients who had received genetic counselling, French genetics health professionals and members of the public. The six genetic counsellors gave an information sheet with the online link to patients who had attended a genetic counselling consultation. An invitation letter, comprising the aim of the study and the link to the survey, was sent to all the members of the French Association of Genetic Counsellors and the French Federation of Human Genetics. People from the general population were recruited in the street by a clinical research technician. Upon establishing contact, one of the two letters and the common survey were randomly drawn at a 1:1 ratio by simple randomisation using an online tool. This questionnaire included four questions about the letter (Table 2) and four questions about characteristics of participants. The primary outcome measure was the preferred letter. An answer of 65% in favour of one of the letters was considered as relevant. With a sample size of 75 participants, the estimation of the expected proportion of 64% (48/75) will have an adequate precision with 95% CI = (53%; 75%). The secondary outcome measure was the proportion of the main feelings experienced after reading letters A and B.

### Statistical analysis

Quantitative variables were described as median (minimum–maximum) and compared using Student's *t* test or the Wilcoxon test according to the distribution of the analysed variable. Qualitative variables were described as frequencies (percentages). Descriptive statistical analyses were performed with a 95% exact binomial confidence interval (CI). Comparisons were performed with the chi-square or Fisher exact test. Statistical analyses were performed using SAS 9.3 (SAS Inc., Cary, NC, USA) software.

## Results

### Study 1: assessment of the impact of the template letter included in the decree (Table 1)

#### Characteristics of the sample

Two groups of participants were recruited, either from genetic consultations ($n = 75$) or from the general population ($n = 73$). The studied sample included 88 females (59.5%) and 60 males (40.5%). The median age was 36 years [18–81]. Sixty-five percent ($n = 97$) had at least one child.

There was no significant statistical difference between the responses from participants from the five recruitment centres, nor between participants' responses and their gender, age and socio-professional category.

#### Understanding

More than 96% ($n/N = 143/148$; 95% CI = [92.3%; 98.9%]) of individuals understood the meaning of the letter (fourth question of questionnaire with response c, d or e). Nevertheless, 27% ($n = 40$) of participants poorly understood some paragraphs, especially the second and fourth paragraphs (P2 and P4), which required a second reading for 38% ($n = 15$) and 35% ($n = 14$) respectively. Seventy-nine percent ($n = 117$) of participants thought that all of the paragraphs in letter A were useful.

#### Feelings

After reading the template letter, the main feeling experienced was anxiety (54%; $n = 80$), while strong anxiety occurred at a lower frequency (8%; $n = 12$). The participants were mainly worried for themselves (54%; $n = 50$). Among worried

**Table 2** Questionnaire of study 3

| | | | Patient | | General pop. | | Professionals | | Total | |
|---|---|---|---|---|---|---|---|---|---|---|
| | | | *N* (76) | % | *N* (75) | % | *N* (80) | % | *N* (231) | % |
| Characteristics of participants | Sex | Female | 48 | 63.2 | 37 | 49.3 | 59 | 73.9 | 144 | 62.3 |
| | | Male | 28 | 36.8 | 38 | 50.7 | 21 | 26.3 | 87 | 37.7 |
| | Age (median) | | 40 | | 36 | | 35 | | 37 | |
| | Had a least one child | | 46 | 60.5 | 39 | 52.0 | 48 | 60.0 | 133 | 57.6 |
| Preference | 1. Which letter do you prefer? | | | | | | | | | |
| | Letter A | | 43 | 56.6 | 35 | 46.7 | 34 | 42.5 | 112 | 48.5 |
| | Letter B | | 33 | 43.4 | 40 | 53.3 | 46 | 57.5 | 119 | 51.5 |
| Understanding | 2. Which letter is more understandable, in your opinion? | | | | | | | | | |
| | Letter A | | 40 | 52.6 | 29 | 38.7 | 34 | 42.5 | 103 | 44.6 |
| | Letter B | | 36 | 47.4 | 46 | 61.3 | 46 | 57.5 | 128 | 55.4 |
| Feelings | 3. After reading the letter, which feelings did you mainly experience? | | | | | | | | | |
| | a. Happy | Letter A | 0 | 0.0 | 0 | 0.0 | 0 | 0.0 | 0 | 0.0 |
| | | Letter B | 0 | 0.0 | 0 | 0.0 | 0 | 0.0 | 0 | 0.0 |
| | b. Serene | Letter A | 5 | 6.6 | 6 | 8.0 | 3 | 3.8 | 14 | 6.1 |
| | | Letter B | 9 | 11.8 | 3 | 4.0 | 5 | 6.3 | 17 | 7.4 |
| | c. Curious | Letter A | 16 | 21.1 | 6 | 8.0 | 11 | 13.8 | 33 | 14.3 |
| | | Letter B | 12 | 15.8 | 12 | 16.0 | 14 | 17.5 | 38 | 16.5 |
| | d. Emotionless | Letter A | 3 | 3.9 | 2 | 2.7 | 6 | 7.5 | 11 | 4.8 |
| | | Letter B | 8 | 10.5 | 8 | 10.7 | 21 | 26.3 | 37 | 16.0 |
| | e. Surprised | Letter A | 19 | 25.0 | 13 | 17.3 | 15 | 18.8 | 47 | 20.3 |
| | | Letter B | 10 | 13.2 | 29 | 38.7 | 14 | 17.5 | 53 | 22.9 |
| | f. Shocked | Letter A | 4 | 5.3 | 12 | 16.0 | 2 | 2.5 | 18 | 7.8 |
| | | Letter B | 3 | 3.9 | 3 | 4.0 | 1 | 1.3 | 7 | 3.0 |
| | g. Worried | Letter A | 25 | 32.9 | 21 | 28.0 | 35 | 43.8 | 81 | 35.1 |
| | | Letter B | 27 | 35.5 | 13 | 17.3 | 23 | 28.8 | 63 | 27.3 |
| | h.Very worried | Letter A | 4 | 5.3 | 13 | 17.3 | 8 | 10.0 | 25 | 10.8 |
| | | Letter B | 7 | 9.2 | 5 | 6.7 | 1 | 1.3 | 13 | 5.6 |
| | i. Angry | Letter A | 0 | 0.0 | 2 | 2.7 | 0 | 0.0 | 2 | 9.0 |
| | | Letter B | 0 | 0.0 | 2 | 2.7 | 1 | 1.3 | 3 | 1.3 |

participants with at least one child ($n = 65$), 49% ($n = 32$) were worried for their children.

Regarding the term "genetic anomaly" used in the proposed letter, 47% ($n = 70$) considered it as appropriate, while 40% ($n = 59$) thought it was frightening. We observed a significant difference depending on the professional background of respondents: those working in the health care sector considered the term "genetic anomaly" to be more anxiety-inducing, compared with other individuals ($p < 0.01$) (Fig. 1). Eighteen percent ($n = 26$) of individuals deemed paragraph P5 as unnecessary. Regarding the sentence in P5 "Some family members may wish to discuss this and others will prefer to remain silent", 53% ($n = 78$) of participants considered it appropriate, while 31% ($n = 46$) believed it was unnecessary. Participants recruited from genetic consultations tended to consider it more appropriate than participants from the general population ($p = 0.07$).

### Intent to contact a health care professional

After reading the letter, 95% ($n = 140$) of individuals reported that they would intend to contact a health care professional if they received the letter personally. This was usually a health care professional associated with a genetic department (50%), but in some cases, a general practitioner (21%) or both (22%).

### Participant's remarks (open-ended questions)

After reading the letter, the main remark of participants was that the letter was clear, well written and adapted. Some thought this "medical information procedure" was acceptable. Some thought the P5 paragraph was inappropriate in this letter because it is not medical, while others (less numerous) found it reassuring. Some felt that this letter was worrying and too

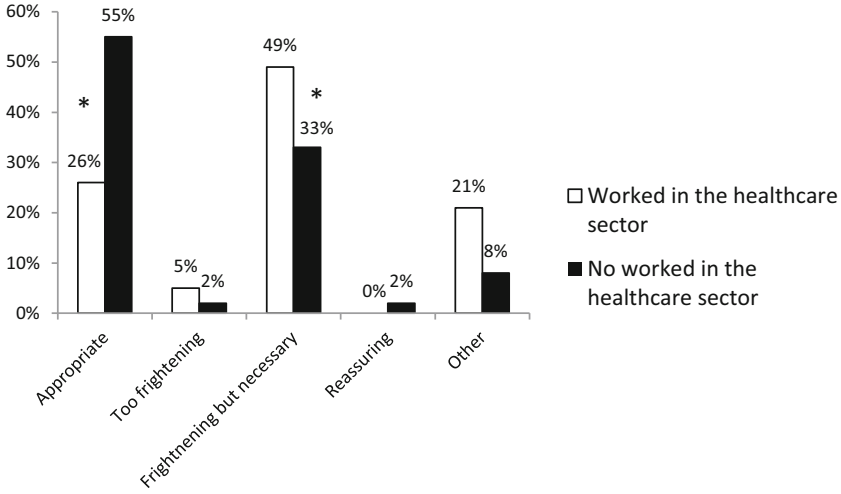


**Fig. 1** Study 1: what do you think about the term "genetic anomaly"? *$p < 0.01$

incentive (feeling of having to see a physician). Some wanted to know the disease in question and its severity.

### Revision of the letter

Based on these results, the focus group wrote a second letter (letter B). The letter A was broadly understood but some paragraphs (P2 and P4) seem complicated. So, the letter B was simpler and shorter. Eighteen percent of individuals deemed paragraph P5 as unnecessary and some thought it was inappropriate in this letter. The focus group removed this paragraph from the letter to judge the relevance of a non-medical paragraph in this letter. Moreover, some genetics centres have written their own template letter; we wanted the letter of the decree to be compared to another.

## Study 2: assessment of the impact of letter B (Table 1)

### Characteristics of the sample

Seventy-five patients and 80 individuals from the general population were recruited. The studied sample included 100 females (64.5%) and 55 males (35.5%). The median age was 40 years [18–80]. Sixty-three percent ($n = 97$) had at least one child.

There were no significant statistical difference between the participants' responses from the five recruitment centres or between participants' responses and their demographic variables.

### Understanding

Ninety-five percent ($n/N = 147/155$; 95% CI = [90.1%; 97.7%]) of individuals understood the meaning of the letter they were given (fourth question of questionnaire with response c, d or e). Only 12% ($n = 18$) of participants poorly understood some paragraphs of letter B, especially the third and second paragraphs, for respectively 44% ($n = 8$) and 39% ($n = 7$), compared with 27% ($n = 40$) after reading letter A ($p < 0.001$). Ninety-four percent ($n = 144$) of participants thought all paragraphs of letter B were useful compared with 79% ($n = 117$) for letter A ($p < .001$).

### Feelings

After reading letter B, the main feeling experienced was anxiety (55%), then curiosity (23%). Among worried respondents who had at least one child ($n = 66$), 68% ($n = 45$) were worried for their children, compared with 49% ($n = 32$) after reading letter A. Very few participants (< 1%) felt anger after reading either letter A or B.

### Intent to contact a health care professional

After reading letter B, 86.5% ($n = 134$) of individuals reported that if they received the letter personally, they would intend to contact a health care professional, usually a health care professional associated with a genetic department (38%), but in some cases, a general practitioner (13%) or both (33.5%).

### Participant's remarks (open-ended questions)

After reading the letter, the main remark of participants was that the letter was clear, concise and well written. Many participants wanted information about the disease, its severity and curability. Some found the letter was too rough, without compassion, and it should be made clear that the participant was not necessarily sick or a carrier. Some participants felt that anonymity was compromised because we know the hospital sending the letter.

### Study 3: comparison between letter A and letter B (Table 2)

#### Characteristics of the sample

Three groups were recruited: patients attending genetic consultations ($n = 76$), individuals from the general population ($n = 75$) and genetic health care professionals ($n = 80$), amounting to 231 participants. The studied sample included 144 females (62%) and 87 males (38%). The median age was 37 years [19–81]. Fifty-eight percent ($n = 133$) had at least one child.

#### Preference

No significant preference for letter A or letter B was found: 48.5% ($n/N = 112/231$; 95% CI = [41.9%; 55.1%]) preferred letter A and 51.5% ($n/N = 119/231$; 95% CI = [44.9%; 58.1%]) letter B. Similarly, there were no significant differences regarding the reading order of the letters or the groups' or participants' demographic variables.

#### Understanding

There was no significant difference between the understanding of the two letters: 45% ($n/N = 103/231$; 95% CI = [38.1%; 51.3%]) of the interviewed individuals thought that letter A was more understandable, while 55% ($n/N = 128/231$; 95% CI = [48.8%; 61.9%]) of participants thought that letter B was easier to comprehend.

#### Feelings

After reading both letters, the main feeling experienced was anxiety (letter A, 46% vs. letter B, 33%), followed by surprise and curiosity. Letter A was considered significantly more worrying than letter B (letter A, 46%; 95% CI = [40%; 52%] vs. letter B, 33%; 95% CI = [27%; 39%], $p = 0.006$). Participants were significantly more relaxed (serene and emotionless participants) after reading letter B than letter A (letter A, 11%; 95% CI = [7%; 15%] vs. letter B, 23%; 95% CI = [18%; 28%], $p = 0.0005$) (Fig. 2).

#### Participant's remarks (open-ended questions)

Some participants preferred the letter A because they found it more human and reassuring, more detailed and clear. Others preferred the letter B because they found it simpler, shorter, clear and concise.

## Discussion

In France, the publication of the decree of 20 June 2013, relating to the article of 7 July 2011 of the French Public Health Code, changed professional practices in the field of clinical genetics. We decided to assess this template letter accompanying this decree, which certain French geneticists and genetic counsellors felt to be inappropriate; for example, paragraph P5 was considered potentially intrusive and anxiety-inducing. However, this concern was not shared by patients or the public, as only a minority of interviewed individuals considered P5 to be unnecessary. It seemed that the majority of patients who had a genetic counselling consultation understood the difficulty of disclosing such information and felt that paragraph P5 was appropriate. Individuals employed in the health care sector considered that the term "genetic anomaly" was more anxiety-inducing compared with other individuals ($p < 0.01$); this may be due to their professional empathy. Study 1 demonstrated a very high level of comprehension of the letter attached to the decree. The most common comments from interviewed individuals about this letter included good formulation/writing of the letter, an acceptable procedure and a necessity to be aware of the disease or at least its severity. Disease characteristics determine the relative's desire to know or not; for example, the desire for the patient to be informed increases slightly depending on whether steps can be taken to prevent the disease and by the gravity of it (Heaton and Chico 2016). However, the French legislator does not allow the mentioning of the name or information concerning the disease in the letter. This could have a negative influence on their intent to contact a HCP, but here, it appears that not because 95% of participants stated that if they received this letter personally they would intend to contact a HCP.

The comparison of letters A and B did not reveal a significant preference for either of the letters. We noted a very good level of comprehension of the two letters. We noted that some paragraphs of letter A seem complicated. However, rephrasing the letter did not interfere with the participants' stated intention to contact a HCP, even if letter B seemed to be more thought-provoking. Anxiety felt by respondents who had at least one child after reading letter B could be explained either by a bias of recruitment or because letter A is considered more human than letter B.

Based on previous literature that indicates genetic information can cause anxiety (Cameron et al. 2009; Catania et al. 2016; Michie et al. 2005), we expected participants to show concern after reading the letters. Both letters were perceived as worrying, however not as worrying as we expected prior to commencing the study; this may be due to the fact that they understood this was a research project and the letters did not apply personally to them.

The aim of the decree and the new "medical information procedure" is to encourage relatives of someone who is

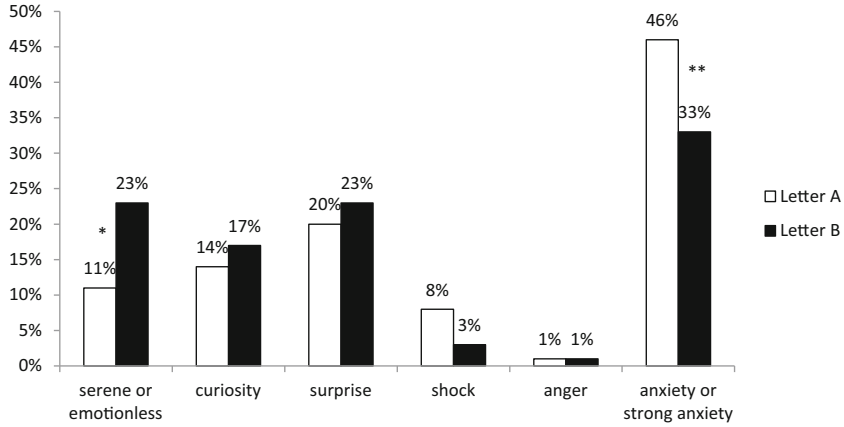


**Fig. 2** Study 3: main reported feelings after reading both letters. *$p = 0.0005$, **$p = 0.006$

affected by a genetic anomaly to make a medical appointment. This study showed that this goal seems to have been achieved, seeing as 95% of participants stated that if they received this letter personally they would intend to contact a HCP. However, we agree that a stated intention does not always equate to actual behaviour, and in clinical practice, some of those who said they would contact a HCP may not do so. In studies 1 and 2, it seemed that the general practitioner (GP) played an important role in this procedure to help families because 43% and 46.5% of participants respectively would contact a GP after reading the letter, emphasising that good collaboration between general practitioners and genetics departments is essential.

No study about the direct sharing of genetic information with relatives by a HCP has already been performed in France. One Australian study (Suthers et al. 2006) and one British study (Kerzin-Storrar et al. 2002) showed that at-risk relatives directly contacted by HCP, with the patient's consent, did not complain of a breach of privacy or autonomy. This intervention improved the family genetic screening. Having a confirmation of hereditary disease and duty to inform its relatives of their risk can be felt as two burdens (Kwiatkowski et al. 2015). The patient may have a poor understanding and think that the HCP is more competent than him to inform his relatives about their risk. At-risk relatives may not become aware of the importance of this information to them and so do not make a medical appointment (Mendes et al. 2017). This "medical information procedure" offers an alternative to patients who do not feel able to inform their at-risk relatives themselves. These Australian and British studies showed the HCP can contact at risk-relatives directly with patient's consent, and that this "medical information procedure" was acceptable to patients and their relatives. This procedure can improve the family genetic screening in France and in other countries if it is applied.

However, it seems important to give priority to direct family communication and to use this approach as a method to increase the proportion of informed at-risk relatives (Mendes et al. 2017). In order to foster communication within families, the HCPs should offer support in the disclosure of genetic information and take family dynamics into consideration. Providing written support about the genetic disease after the consultation to assist the individual with the communication process helps increase the at-risk relative's level of awareness (Forrest et al. 2003), and the number of at-risk relatives having appointments with a genetics service (Forrest et al. 2008; Gorrie et al. 2017; Kerzin-Storrar et al. 2002;Wright et al. 2002).

Before the legislative order dated 8 December 2014, the responsibility of communicating the genetic information to families was unclear. By introducing the decree, the legislator relieves the HCP of this responsibility. After the HCP has informed the patient about any risks to relatives, it is the patient themselves who engage their liability to inform them and risk conviction for offences by a judge. The decree delivers a strong message regarding health management to modify relationships between HCPs and patients. This medical consultation involves a medical and legal relationship, whereby the HCP has to inform patients about their responsibility and the risks. This point creates some discomfort in the French medical community, as the medical relationship with the patients may be altered because of mutual medical and legal protection.

## Study limitations

The most relevant bias was that participants had to respond to a hypothetical situation. Responses to a real procedure could be different and should be assessed. It was chosen to study the general population because it represents at best the individuals who could receive this letter. These are individuals who read a letter about their genetic risk while they have never had information about genetics in consultation. We wanted to compare their responses with those of patients who had a genetic counselling consultation. The participants of studies 1 and 2 were different, which could explain the differences in responses in the results of both studies. We cannot exclude some selection bias of the study populations. However, the

randomised part of our assessment (study 3) ensured an unbiased comparison of templates A and B.

## Conclusion

Health care professionals must consider the difficulties for individuals to communicate the genetic risk to their relatives and help them in this process, for example, by providing written support that individuals could use and give to their relatives to conform to the decree of June 2013.

Given that our study did not show any major differences between understanding and anxiety after the two letters, every HCP can decide whether to use the letter of the decree or adapt this letter as they see fit.

Follow-up research could assess the rate of relatives having received this letter who have appointments with a genetics service. Further research could assess the acceptance and the application of this familial information procedure by genetic professionals.

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### Compliance with ethical standards

All procedures followed were in accordance with the ethical standards. This study was approved by the Research Ethics Committee (CPP SOOM III: Comité de Protection des Personnes Sud-Ouest et Outre-Mer III). Informed consent was obtained from all participants included in this study.

**Conflict of interest** The authors declare that they have no conflict of interest.

## References

American Society of Human Genetics , Social Issues Subcommittee on Familial Disclosure(1998), Professional disclosure of familial genetic information. Am J Hum Genet 62(2):474–483 at 474

Base SAS® 9.3 Procedures Guide Copyright © 2011, SAS Institute Inc., Cary, NC, USA

Cal (1976) Text of Tarasoff v. Regents of the University of California, 17 Cal. 3d 425, 551 P.2d 334, 131 Cal. Rptr. 14

Cameron LD, Sherman KA, Marteau TM, Brown PM (2009) Impact of genetic risk information and type of disease on perceived risk, anticipated affect, and expected consequences of genetic tests. Health Psychol 28(3):307–316. https://doi.org/10.1037/a0013947

Catania C, Feroce I, Barile M, Goldhirsch A, De Pas T, de Braud F, Boselli S, Adamoli L, Radice D, Rossi A, Spitaleri G, Noberasco C, Bonanni B (2016) Improved health perception after genetic counselling for women at high risk of breast and/or ovarian cancer: construction of new questionnaires - an Italian explory study. J Cancer Res Clin Oncol 142(3):633–648. https://doi.org/10.1007/s00432-015-2062-7

Clarke A, Martin R, Kerzin-Storrar L, Halliday J, Youg MA, Simpson SA, Featherstone K, Foorest K, Lucassen A, Morrison PJ, Quarrekk OWJ, Stewart H, Collaborators (2005) Genetic professionals' reports of nondisclosure of genetic risk information within families. Eur J Hum Genet 13(5):556–562

d'Audiffret D, de Montgolfier S (2016) Pratiques professionnelles et enjeux éthiques associés à l'ajout du conseil génétique dans la loi sur l'information à la parentèle en génétique humaines ?. 8èmes Assises de Génétique Humaine et Médicale. Lyon. 3-5/02/2016

Dheensa S, Fenwick A, Shkedi-Rafid S, Crawford G, Lucassen A (2016) Health-care professionals' responsibility to patients' relatives in genetic medicine: a systematic review and synthesis of empirical research. Genet Med 18(4):290–301

Forrest K, Simpson SA, Wilson BJ, van Teijlingen ER, McKee L, Haites N, Matthews E (2003) To tell or not to tell: barriers and facilitators in family communication about genetic risk. Clin Genet 64(4):317–326

Forrest LE, Delatycki MB, Skene L, Aitken M (2007) Communicating genetic information in families–a review of guidelines and position papers. Eur J Hum Genet 15(6):612–618

Forrest LE, Burke J, Bacic S, Amor DJ (2008) Increased genetic counseling support improves communication of genetic information in families. Genet Med 10(3):167–172

Forrest LE, Delatycki MB, Curnow L, Skene L, Aitken MA (2010) Genetic health professionals and the communication of genetic information in families: practice during and after a genetic consultation. Am J Med Genet A 152A(6):1458–1466

Gorrie A, Archibald AD, Ioannou L, Curnow L, McClaren B (2017) Exploring approaches to facilitate family communication of genetic risk information after cystic fibrosis population carrier screening. J Community Genet 9(1):71–80. https://doi.org/10.1007/s12687-017-0337-1

Heaton TJ, Chico V (2016) Attitudes towards the sharing of genetic information with at-risk relatives: results of a quantitative survey. Hum Genet 135(1):109–120

Kerzin-Storrar L, Wright C, Williamson PR, Fryer A, Njindou A, Quarrell O, Donnai D, Craufurd D (2002) Comparison of genetic services with and without genetic registers: access and attitudes to genetic counselling services among relatives of genetic clinic patients. J Med Genet 39(12):e85

Kwiatkowski F, Laquet C, Dessenne P, Bignon YJ (2015) Informer la famille: émotions et attitudes du consultant en oncogénétique pour risque familial de cancer sein/ovaire ou côlon. Bull Cancer 102(2):162–173. https://doi.org/10.1016/j.bulcan.2014.09.001

Lahlou-Laforet K, Albuisson J, Mazzella JM, Moliere D, Laurent-Puig P, Jeunemaitre X, Consoli S, Gimenez-Roqueplo AP (2014) Décret du 20 juin 2013 relatif à la procédure d'information aux apparentés. Appliquer la loi sans traumatiser la parentèle : analyse psychologique et propositions pratiques. 7èmes Assises de Génétique Humaine et Médicale. Bordeaux. 29–31/01/2014

Law no.2004–800 dated of 6 August 2004 - art. 4. (2004). Code de la santé publique. L1131-1. JORF

Law no.2011–814 dated of 7 July 2011 - art. 2. (2011). Code de la santé publique. L1131-1. JORF

Legislative Decree 2013-527 20th June 2013 concerning the implementation of conditions of kin information in the context of an examination of genetic characteristics for medical purposes. R. 1131-20-2 du code de la santé publique. JORF n°0143, 22 June 2013, p10403

Legislative order fixing the sample letter addressed by the physician to potentially affected members of the family pursant to Article R.

1131-20-2 du code de la santé publique. JORF n°0143, 22 June 2013, p10405

Legislative order defining the rules of good practice applicable to the implementation of kinship of information in the context of an examination of genetic characteristics for medical purposes pursant to Article R. 1131-20-2 du code de la santé publique. JORF n°0293, 19 December 2014, p21495

Mendes A, Metcalfe A, Paneque M, Sousa L, Clarcke AJ, Sequeiros J (2017) Communication of information about genetic risks: putting families at the center. Fam Process 57:836–846. https://doi.org/10.1111/famp.12306

Michie S, Lester K, Pinto J, Marteau TM (2005) Communicating risk information in genetic counseling: an observation study. Health Educ Behav 32(5):589–598

Nycum G, Avard D, Knoppers BM (2009) Factors influencing intrafamilial communication of hereditary breast and ovarian cancer genetic information. Eur J Hum Genet 17(7):872–880

Parker M, Lucassen A (2003) Concern for families and individuals in clinical genetics. J Med Ethics 29(2):70–73

Suthers GK, Armstrong J, McCormack J, Trott D (2006) Letting the family know: balancing ethics and effectiveness when notifying relatives about genetic testing for a familial disorder. J Med Genet 43(8):665–670

Wright C, Kerzin-Storrar L, Williamson PR, Fryer A, Njindou A, Quarrell O, Donnai D, Craufurd D (2002) Comparison of genetic services with and without genetic registers: knowledge, adjustment, and attitudes about genetic counselling among probands referred to three genetic clinics. J Med Genet 39(12):e84