Original Article

PORTO BIOMEDICAL JOURNAL
WHERE SCIENCE MEETS KNOWLEDGE

OPEN

# Identifiable relatives in the family history: not without individual consent

José Pedro L. Nunes, MD[a,*], Maria do Sameiro Faria, MD[b], Carlos Abreu Amorim, PhD[c]

**Abstract**

The family history is a traditional section of the clinical record. Data on family members in the clinical record may be anonymous but yet these may be easily identifiable; therefore, exposing the relatives of the patient to the fact that a written record is produced, mentioning them, without their consent. This is in direct contradiction with European data protection and other regulations and in contradiction with a reasonable ethical perspective. For the purpose of obtaining an image of the present state of affairs, we used as a convenience sample, the series of Case Records published in 2019 in *The New England Journal of Medicine* (January to December). From a total number of 40 reports, identifiable relatives were present in 30. The number of identifiable relatives varied between none and 6. It is not the right of each individual to disclose sensitive clinical information regarding other persons, without consent from these latter. Family history should no longer include identifiable relatives, unless consent is obtained from each identifiable person. The authors offer the following guidelines on this topic: (1) Do not mention any identifiable relative of the patient in the medical history without consent from the said relative; (2) Do not mention in the family history clinical conditions seemingly unrelated to the present clinical situation; (3) Do not mention in the family history clinical conditions that the patient does not (him/) herself have and that may be seen as social stigmata; (4) Consult the institutional Ethics committee in case of reasonable doubt.

**Keywords:** consent, data protection, family history

## Introduction

The family history is a traditional part of the clinical record. Because many diseases show an increased incidence in patients with relatives with the same disease, questioning about the family history is an easy way to evaluate the probability that a given disease may in fact exist in a given patient, at least in some cases. It is also clear that the genetic similarity tends to be greater, the more a closer relation of kinship exists. This means that if a first-degree relative has a medical condition, this will usually be more relevant than if a distant cousin has the same disease.

In Europe, recent regulations[1] have changed the way data concerning each and every individual should be treated, indicating that consent must be obtained from any person to be mentioned in recorded data. Elsewhere in the world, legislation has been changing in a similar direction.[2]

The problem at hand, to be analyzed in the present report, is that people mentioned in a clinical record may be anonymous but yet be easily identifiable; therefore, exposing the relatives of the patient to the fact that a written record is produced, mentioning them, without their consent – in contradiction with European and other regulations and in contradiction with a reasonable ethical perspective.

## Case study

For the purpose of obtaining an image of the present state of affairs, we used as a convenience sample, the series of Case Records published in 2019 in *The New England Journal of Medicine* (January to December; Table 1). The following data were retrieved from each report: age, sex, diagnosis, family history. In the case that any identifiable relative was mentioned in the text, the number of such persons was counted.

From a total number of 40 reports, identifiable relatives were present in 30. The number of identifiable relatives varied between none and 6 (Table 1).

Identifiable relatives, presented in an anonymous way, were present in a considerable fraction of the reports studied (although, in a number of the cases presented, the fact of being identifiable may be debatable). As these were taken as a convenience sample, they may not be representative of the situation in clinical practice, neither in the USA nor in other countries. Furthermore, the published texts may hold a considerable degree of deviation from the actual original and corresponding clinical records.

### *The European general data protection regulation*

Published in 2016 and enforced in 2018, the European Union General Data Protection Regulation[1] lays down some principles concerning the issue at hand:

1. "The principles of data protection should apply to any information concerning an identified or identifiable natural person"
2. "Consent should be given by a clear affirmative act establishing a freely given, specific, informed, and unambiguous

*Sponsorships or competing interests that may be relevant to content are disclosed at the end of this article.*

*[a] Faculdade de Medicina da Universidade do Porto, [b] Centro Materno Infantil do Norte, Porto, [c] Escola de Direito da Universidade do Minho, Braga, Portugal.*

*[*] Corresponding author. Faculdade de Medicina da Universidade do Porto, Porto, Portugal. E-mail address: jplnunes@med.up.pt (José Pedro L. Nunes).*



*Porto Biomed. J. (2020) 5:2(e61)*

*Received: 29 January 2020 / Accepted: 5 February 2020*

*http://dx.doi.org/10.1097/j.pbj.0000000000000061*

**Table 1**

**Case records published at *The New England Journal of Medicine* in 2019 (January–December), in what regards age, sex, main diagnosis, and details of the family history**

| Case number | Age (yr)/sex | Diagnosis | Family history | Identifiable relatives with medical condition(s) |
|---|---|---|---|---|
| 1 | 34/Male | Posttraumatic stress disorder | Father/depression; mother, father, and paternal uncle/alcohol and drug use disorders; sister died from bone cancer | Yes (4) |
| 2 | 36/Male | Cutaneous tuberculosis | Father/died with emphysema | Yes (1) |
| 3 | 70/Female | Powassan virus encephalitis | Family history/Alzheimer disease, stroke | No |
| 4 | 18/Male | Perforation of the sigmoid colon by a foreign body (toothpick) that caused a fistula to the right common iliac artery | No family history of autoimmune diseases or inflammatory bowel disease | No |
| 5 | 48/Female | Pernicious anemia | Family psychiatric and medical history unknown; patient adopted | No |
| 6 | 29/Female | Oxycodone and cocaine use | Family history unknown; patient adopted | No |
| 7 | 73/Female | Herpes simplex virus lymphadenitis Chronic lymphocytic leukemia | Mother/heart disease; father/skin cancer | Yes (2) |
| 8 | 58/Female | Ocular syphilis | Father/Crohn disease; mother/glaucoma, breast cancer; 1 sister/Hashimoto thyroiditis; other sister/endometrial cancer | Yes (4) |
| 9 | 62/Male | Symptomatic atrial fibrillation with associated anxiety | Wife/died with cancer; girlfriend/recurrent cancer; parents/heavy alcohol use; several paternal relatives/ depression; maternal aunt/died of an intentional drug overdose; mother/emphysema, died of breast cancer; father/ hyperlipidemia, died of asbestosis | Yes (5) |
| 10 | 69/Male | Idiopathic pulmonary fibrosis | Father/died of myocardial infarction; mother/basal cell carcinoma; no family history of lung or autoimmune disease; 4 children/healthy | Yes (6) |
| 11 | 49/Male | End-stage renal disease in a patient with human immunodeficiency virus infection | Family history of diabetes mellitus, coronary artery disease, and blood clots | No |
| 12 | 60/Male | Motor neuron disease with TAR DNA-binding protein 43 proteinopathy. Amyotrophic lateral sclerosis. Acute bronchopneumonia | No family history of neurologic disease | No |
| 13 | 54/Male | Surreptitious ingestion of isopropyl alcohol | Parents, 2 brothers/alcohol use disorder | Yes (4) |
| 14 | 44/Male | Metastatic postpubertal immature teratoma | No family history of coronary artery disease or cardiomyopathy. Mother/hypertension, diabetes; father, brother, 2 children healthy | Yes (1) |
| 15 | 55/Male | Acute hepatitis B virus and hepatitis delta virus coinfection in the presence of chronic hepatitis C virus infection | Parents deceased; medical history of siblings unknown | No |
| 16 | 53/Male | Tropical pulmonary eosinophilia | Several first-degree relatives/hypertrophic cardiomyopathy; mother/died from pulmonary tuberculosis | Yes (1) |
| 17 | 44/Male | Systemic lupus erythematosus | One son/symptoms of an upper respiratory infection; no family history of heart disease, human immunodeficiency virus infection, tuberculosis, autoimmune disease, or cancer | Yes (1) |
| 18 | 24/Female | Anaplastic carcinoma arising in association with intestinal-type mucinous carcinoma of the ovary | Paternal grandmother/uterine cancer; paternal grandfather/ gastric and prostate cancer; paternal aunt/breast cancer; paternal uncle/prostate cancer; maternal grandmother/died of liver cancer associated with hepatitis C virus; parents, sister/healthy | Yes (5) |
| 19 | 38/Female | Intestinal tuberculosis | Mother/coronary artery disease; father/diabetes and hypertension | Yes (2) |
| 20 | 52/Female | Chronic Chagas disease with reactivation of latent *Trypanosoma cruzi* infection | Father/died from trauma; mother/died from kidney disease; no family history of coronary artery disease, cardiomyopathy, or sudden cardiac death | Yes (2) |
| 21 | 31/Female | Leber hereditary optic neuropathy | Grandfather/hyperlipidemia; mother/glaucoma; no other family members had a history of vision loss | Yes (2) |
| 22 | 65/Female | Statin-associated autoimmune myopathy | Father/died of myocardial infarction; mother/hypertension, osteoarthritis, hip replacement; brother/prostate cancer; sister/sarcoidosis | Yes (4) |
| 23 | 52/Male | Flail mitral valve due to acute myocardial infarction of the papillary muscle in the absence of obstructive coronary artery disease | Father/died of myocardial infarction; 2 paternal uncles/died of coronary artery disease | Yes (3) |
| 24 | 39/Female | Hyperthyroidism due to Graves disease | No medical problems in the family reported | No |

(*continued*)

**Table 1**
**(continued).**

| Case number | Age (yr)/sex | Diagnosis | Family history | Identifiable relatives with medical condition(s) |
|---|---|---|---|---|
| 25 | 41/Female | Acute suppurative appendicitis and periappendicitis | Father/died of liver disease associated with alcohol use disorder; mother/cervical cancer, hypertension, died of a ruptured cerebral aneurysm | Yes (2) |
| 26 | 27/Female | Opioid use disorder and malingering | Multiple relatives, including both parents/substance use disorder | Yes (2) |
| 27 | 16/Female | Concussion, benign paroxysmal positional vertigo, and attention deficit hyperactivity disorder | Brother/attention deficit hyperactivity disorder; no family history of headaches, learning disabilities, depression, anxiety, or seizure disorders | Yes (1) |
| 28 | 22/Female | Interstitial lung disease associated with FLNA mutation | Younger brother/seizure disorder and developmental delays; mother/mild joint hypermobility; father/healthy | Yes (2) |
| 29 | 1/Male | Congenital esophageal stenosis with fibromuscular thickening of the esophagus | Mother/anemia; father, sister/healthy; maternal aunt/ hyperthyroidism; maternal grandmother/pancreatitis; maternal grandfather/hypertension | Yes (4) |
| 30 | 65/Female | Active myocarditis consistent with myocarditis related to immune checkpoint inhibition | Father/coronary artery disease; mother/ lung cancer, osteoporosis, spinal stenosis | Yes (2) |
| 31 | 45/Female | Adenovirus (serotype 2) meningoencephalitis | Mother/coronary artery disease, diabetes, and migraine; father/ hypertension, diabetes, atrial fibrillation, multiple sclerosis | Yes (2) |
| 32 | 70/Female | Creutzfeldt-Jakob disease | Mother/hypertension; father/died from lung cancer; brother, 2 adult children healthy; no family history of ataxia, dementia, autoimmune disease, or neurodegenerative disease | Yes (2) |
| 33 | 35/Female | Amniotic fluid embolism | Husband/human immunodeficiency virus type 1 (HIV-1) infection; unknown family medical history | Yes (1) |
| 34 | 16/Male | B-cell acute lymphoblastic leukemia | Mother/fatigue, jaundice, abnormally elevated results of liver function tests, spontaneous resolution; maternal grandfather/nonalcoholic fatty liver disease, brain cancer; maternal second cousin/systemic lupus erythematosus | Yes (2) |
| 35 | 66/Male | Skin involvement associated with peripheral T-cell lymphoma | Father/died from coronary artery disease; brother/sarcoidosis and alpha1-antitrypsin deficiency; aunt and uncle/cancer | Yes (4) |
| 36 | 34/Male | Kaposi sarcoma of the gastrointestinal tract | None reported | No |
| 37 | 1.7/Male | Juvenile myelomonocytic leukemia | Father/multiple café au lait macules; mother and father/ substance use disorders | Yes (2) |
| 38 | 20/Male | Diffuse pulmonary alveolar hemorrhage with focal infarction, multifocal thromboembolic disease, and neutrophilic infiltrate | Father/obstructive sleep apnea; paternal grandfather/chronic obstructive pulmonary disease; maternal grandfather/ rheumatoid arthritis, possible pulmonary fibrosis; 3 maternal cousins/ multiple sclerosis | Yes (6) |
| 39 | 57/Female | Control of hemorrhage with the use of resuscitative endovascular balloon occlusion of the aorta | None reported | No |
| 40 | 26/Female | Lymphocytic choriomeningitis virus infection | Mother, father/healthy | No |

Presented are the presence and number of identifiable relatives mentioned in each text. Original texts available at https://www.nejm.org/.
FLNA = filamin A.

indication of the data subject's agreement to the processing of personal data relating to him or her, such as by a written statement, including by electronic means, or an oral statement"
3. "Personal data which are, by their nature, particularly sensitive in relation to fundamental rights and freedoms merit specific protection"
4. "The data subject be informed of the existence of the processing operation and its purposes"
5. "Processing shall be lawful only if and to the extent that at least one of the following applies: (a) the data subject has given consent to the processing of his or her personal data for one or more specific purposes; (b) processing is necessary to protect the vital interests of the data subject or of another natural person"

### *The California consumer privacy act*

In the USA, there is no text of legislative nature similar to the European Regulation. Among the relevant legislative texts stands the California Consumer Privacy Act.[2] Published in 2018 and enforced in January 2020, the Act recognizes the right of privacy, and further states that "Fundamental to this right of privacy is the ability of individuals to control the use, including the sale, of their personal information." Similarly to the European counterpart, this bill also holds the following points:

1. "require a business to make disclosures about the information and the purposes for which it is used";
2. "grant a consumer the right to request deletion of personal information";
3. "require the business to delete upon receipt of a verified request."

### *Brazilian data protection regulation*

Similarly to the situation in Europe and in California, also in Brazil (República Federativa do Brasil) a data protection regulation[3] has been published (in 2018; enforced in 2020).

The Brazilian regulation follows some aspects of the European Union counterpart, and although exceptions exist, the general rule is that "o tratamento de dados pessoais sensíveis somente poderá ocorrer nas seguintes hipóteses: I - quando o titular ou seu representante legal consentir, de forma específica e destacada, para finalidades específicas" (The processing of sensitive personal data may only occur under the following circumstances: I – when the holder or his/her legal representative specifically and prominently consents, for specific purposes).

Furthermore, personal data are defined as being "information related to an identified or identifiable natural person."

### *Identifiable relatives in the family history and data protection*

Clinical information of a sensitive nature is frequently present in the family history. Because in many cases the relatives mentioned in the family history are readily identifiable – as in the case of a spouse – we are not dealing with truly anonymized data. In our view, informed consent, from the part of each identifiable relative, should have been obtained before that information being recorded.

It is not the right of each individual to disclose sensitive clinical information regarding other persons, to help the doctors in charge of him/herself.

Some of the Case Records analyzed presented the family history in a manner compatible with the present text. For instance, Zachary et al[4] presented the data in the following way: "There was a family history of Alzheimer's disease and stroke." King et al[5] present no family history, and the text ends with an interesting "patient perspective."

Andreasen et al[6] presented, as an alternative to the classic family history method, the family study method, consisting in interviewing directly relatives. This approach can overcome some difficulties seen with the classic method.

Concerning the European Regulation mentioned above, the text does admit an exception, whenever "processing is necessary to protect the vital interests of the data subject or of another natural person." We are unware of any evidence demonstrating that mentioning the concrete persons, in the family history, further protects the "vital interests" of the "data subject" (identifiable relative) as an alternative, namely, to a general mention with no reference to concrete persons. We therefore reject that references to identifiable relatives in the family history may be made in accordance with the European Regulation[1] under discussion.

It may further be argued that the family history need not contain information seemingly unrelated to the present clinical situation.

Concerning diseases that represent social stigmata, it may be argued that such data not be included in the family history, as long as the situation does not affect the patient him/(her)self. In the report by Bernstein et al,[7] it is stated that the patient's husband had human immunodeficiency virus type 1 infection. It would have been possible to withhold this information, perhaps by stating something similar to "the patient had previously maintained close contact with a person with human immunodeficiency virus type 1 infection," if deemed strictly necessary.

**Table 2**

**Suggestion for guidelines to be followed concerning identifiable relatives and the family history**

| |
|---|
| 1. Do not mention any identifiable relative of the patient in the medical history without consent from the said relative. |
| 2. Do not mention in the family history clinical conditions seemingly unrelated to the present clinical situation. |
| 3. Do not mention in the family history clinical conditions that the patient does not (him/) herself have and that may be seen as social stigmata. |
| 4. Consult the institutional ethics committee in case of reasonable doubt. |

### *Suggested guidelines*

We offer a suggestion for guidelines to be followed concerning identifiable relatives and the family history in Table 2.

## Conclusions

It is not the right of each individual to disclose sensitive clinical information regarding other persons, without consent from these latter. Family history should no longer include identifiable relatives, unless consent is obtained from each identifiable person.

## Acknowledgments

None.

## Author contributions

J.P.L.N. planned the study and wrote the draft. J.P.L.N. and M.S.F. retrieved the data presented in Table 1. J.P.L.N. and C.A.A. reviewed the regulatory and legal issues. J.P.L.N., M.S.F., and C.A.A. revised the text for critical intellectual content. J.P.L.N. submitted the paper.

## Conflicts of interest

The authors declare no conflicts of interest.

## References

[1] European Union. Regulation (EU) 2016/679 of the European Parliament and of the Council of 27 April 2016 on the protection of natural persons with regard to the processing of personal data and on the free movement of such data, and repealing Directive 95/46/EC (General Data Protection Regulation). Official Journal of the European Union. Volume 59. L 119/1 - L119/88. 4 May 2016.

[2] State of California. Assembly Bill No. 375. Privacy: personal information: businesses. 2018. (amendment to the California Civil Code).

[3] Senado Federal (República Federativa do Brasil). Lei n° 13.709 de 14/08/2018 (Lei geral de proteção de dados). Diário Oficial da União, N° 157, pages 59-64. 2018.

[4] Zachary KC, LaRocque RC, Gonzalez RG, Branda JA. Case 3-2019: a 70-Year-Old Woman with Fever, Headache, and Progressive Encephalopathy. N Engl J Med. 2019;380:380–387.

[5] King DR, Crowley JC, Frenk NE, Kaafarani HMA. Case 39-2019: a 57-year-old woman with hypotension and trauma after a motorcycle accident. N Engl J Med. 2019;381:2462–2469.

[6] Andreasen NC, Endicott J, Spitzer RL, Winokur G. The family history method using diagnostic criteria: reliability and validity. Arch Gen Psychiatry. 1977;34:1229–1235.

[7] Bernstein SN, Cudemus-Deseda GA, Ortiz VE, Goodman A, Jassar AS. Case 33-2019: a 35-year-old woman with cardiopulmonary arrest during cesarean section. N Engl J Med. 2019;381:1664–1673.