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Information Assessment for the Implementation of Electronic Informed Consent for Genetic Studies in a High Complexity Hospital

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Abstract

The objective of this study was to investigate and analyze the most relevant aspects that influence the development and implementation of electronic informed consent for genetic studies. Interviews were conducted with experts in the area within our institution, the different informed consents available and the number of genetic studies requested in the last 5 years were analyzed.

Professionals acknowledged the ethical dilemmas related to the genetic studies and the importance of having an electronic informed consent that not only provides the patient with the information necessary to understand the implications of the study, but also be flexible enough to adapt to the various genetic studies today.

The development of informed consent is a challenge for health IT professionals, due to the complexity of the information it contains and the ethical implications it represents.

Keywords:

Genetic test, informed consent, electronic health record.

Introduction

The increasing use of genetic/genomic information is an important aspect of modern medicine. Sequencing technologies are being refined, allowing greater amounts of genomic data to be obtained in shorter times. These advances make it possible to have a growing volume of raw material to carry out studies on genomic data and generate more knowledge about the relationship between genomics and health.

Several tests are ordered for health institutions year after year. In 2018, Phillips et al, reported 75,000 genetic tests on the market in the USA, approximately. Prenatal tests represent the highest percentage of spending on genetic tests, and hereditary cancer tests accounted for the second-highest [1]. In Argentina, the amount of genetic testing currently performed is difficult to estimate accurately. Penchaszadeh reported in 2013, 7,700 non-prenatal genetic studies conducted during the period 1998 to 2008 in a public children's hospital. However, it clarifies that reports belonging to the Argentine private health sector are scarce or unknown [2].

Genetic/genomic studies have become increasingly integrated into clinical research and patient care. This genetic information begins to increase in the electronic medical record and raises concerns about privacy and data protection in the EHR.

Due to the aforementioned, the main ethical challenges linked to these types of studies include those related to privacy, data protection, insurance, public-private collaboration, genetic discrimination and the management of unforeseen outcomes whose clinical importance is uncertain as explained by Minari et al in their 2018 publication [3].

With regard to data protection, information technologies have a key role ensuring high security in the use, storage and exchange of genetic data, in this way the risk of violation of confidentiality along with other patient rights can be minimized. These rights along with those that contemplate genetic heritage are promoted and protected in our country by several laws. Among them we can mention the law 712 of the city of Buenos Aires, "Law on Guarantees of Genetic Heritage" and the National Law 25.326 "Protection of Personal Data."

Although we have previously mentioned relevant aspects of genetic studies, it is important to emphasize that prior to the performance of such tests, informed consent is required in the first instance. Consent is not only useful as a legal basis for the processing of this type of data but also, it is the means by which the patient understands the procedure to be performed, the benefits and their rights over the results. Currently, electronic informed consents have proven to be of greater understanding for patients than those on paper [4].

Electronic informed consent for genetic studies is a major challenge for information systems, due to the complexity of the information they contain, the lack of guidelines to guide their development and the ethical challenges they entail.

In this paper we describe the process of analysis and survey carried out within our institution with the aim of computerizing the informed consent for generic studies within the Information System of the Hospital Italiano. For this purpose, interviews were conducted with experts on relevant topics in the literature, consultations with the area of statistics on genetic studies and a collection of informed consents in use.

Methods

Settings

The Hospital Italiano (HIBA) is a high complexity medical center located in the City of Buenos Aires. HIBA is a HIMSS Stage 7 organization with an in-house-developed health information system. It features a web-based, problem-oriented EHR; a terminology server referenced to SNOMED CT; and an integrated personal health record (PHR) [5].

More than 7,000 genetic tests were prescribed during the last two years. The number is increasing annually according to internal records. Some of the most requested tests were Prothrombin Mutation G20210A, CD34 Marker, JAK-2 V617F Mutation and PML RAR Alpha (PCR), among others. Main solicitor services were Hematology, Clinical Medicine, Transfusion Medicine, Neurology, Pediatrics, Reproduction - Gynecology and Family Medicine.

The institution uses Ion Reporter (Thermo Fisher) software to process and analyze genomic data generated by Ion Torrent sequencers. It has recently developed an in-house application for filtering genomic variants in order to improve the identification of relevant variants process: LabSec. It can be comparable to commercial software Sophia Genetics DDM or Agilent Cartagenia among others, but according to the

particular needs of our users in terms of visibility and data management. It allows managing a great amount of data through a user-friendly interface, reducing errors and streamlining in the filtering process.

Interviews with professionals

We conducted open interviews, both individual and group, with referents of the area within the Hospital Italiano. Among them we can highlight: safety and genomics committees, the standards and procedures area of the institution, a group of specialists in hereditary cancer in adults (composed of doctors specialized in colon, gynecology, dermatology and other areas) and different professionals in the area of Pediatrics.

The interviews were conducted over the course of one year, between August 2019 and September 2020. In total, 8 meetings were held with the different professionals.

As prompts for the interviews, relevant topics were collected from the literature. For them, we focus on ethical points, gray areas of regulation and legal aspects. The main themes highlighted were:

- Results involving biological family members
- Incidental findings
- Results including variants of uncertain significance
- Scientific research
- Particular needs of professional practice

In addition, professionals were consulted on the different informed consents in use during their practice and current examples were collected.

Results

There is general agreement among professionals as to the importance and need for computerization of informed consents in use. Currently, we find a diversity of paper documents being used. 6 current informed consents were obtained from the interviewees. Of these, 4 correspond to different external genetic laboratories, while the remaining two were written in the HIBA.

One of the most obvious points of the interviews was the difference in needs between the different domains and types of study. Professionals were interested in having computerized consents meet the particular needs of their practice. We detected as main categories to differentiate germinal and somatic practices, as well as whether the study is a panel of genes, an exome or the entire genome of the patient.

As for the way in which the patient can express his will by completing the consent, there was no general agreement on the possibility that the patient can select from several options. However, the view to keep simple options limited to “yes” or “no” was highlighted.

On the incidental findings, there was total agreement on informing patients. This position is in line with the recommendations in Argentina. However, in the face of variants of uncertain significance , the results were mixed, with the professional criteria, the relevance of the finding and the quality of the study taking precedence to make the decision to communicate them.

The professionals recognized the ethical dilemmas related to findings with possible impact on biological relatives. Recommendations were aimed at protecting the privacy of their patients. The importance of a regulatory framework to address this issue was identified. Currently, they communicate to the patient the possibility of impact on their direct relatives, leaving to them the decision to inform their relatives of the findings or recommend genetic consultations.

Regarding the ability to see genetic results in the EHR there was no general agreement among the interviewees. The two main positions were: only geneticists can have access to them (arguing that they are the most trained to understand and interpret them); all medical professionals could see them (considering the importance of not hiding information within the clinical record). Currently, HIBA does not limit the ability to see genetic results by role or area of the medical professional.

On the patient side, on the ability to see results in the patient portal (PHR) the recommendation was to use a model similar to HIV results: enable the patient to see results once the patient was informed by the doctor. In this way, the patient always receives the interpretation of the result at the genetic consultation.

A common point among all interviewees is the value of genetic consultation both before and after the completion of the studies. Most interviewees recommended that only genetic specialists be able to prescribe and interpret these studies.

Discussion

It stands out from the interviews that the dilemmas and problems related to genetic studies referred by HIBA professionals are in line with those described in the literature.

The diversity in the origin of different consents in use can be a challenge to the success of the project. However, the interest of professionals in computerization makes the line of work relevant. The content of already existing documents can be a firm basis for the creation of informed consent for the institution.

Within the areas of common agreement included the report of the variants of uncertain significance, the value of genetic counseling in patient care, the difference between the different domains (somatic vs germinal) and the different types of study (panel, exome and genome), as well as limiting the display in the PHR by the patient before the medical interview.

Privileges for the prescription of studies and visualization of results in the EHR is an area where consensus must still be built, so they must be treated with caution within the project.

It is evident that there is a need to incorporate different areas outside of medical professionals in the decision - such as the legal area and the Ethics Committee, since there are plenty of high-impact areas that need their action and knowledge.

Conclusions

This paper describes the survey carried out as the first stage for the implementation of a computerized informed consent for genetic/genomic studies in the EHR. The results obtained are valuable for the design and implementation of a solution that includes all the use cases and needs of professionals and patients. There is a clear need for informed consent with sufficient flexibility to suit all professionals. Work will continue to seek consensus to promote the success of the project.

A secondary benefit of this experience was the creation of networks and open communication spaces between genetics professionals and the health informatics team in charge of the project.

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