Privacy Considerations in Medical Publications – A Rare Perspective

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Abstract

Aim: Individual privacy has come under increasing scrutiny since the passage of the General Data Protection Regulation. While patient privacy is crucial in all publications, the challenge is particularly severe in rare diseases where, by definition, there are very few patients, thereby increasing the risk of identification.
Methods: We searched PubMed and Google for articles on patient privacy in rare diseases. Search terms included "rare" OR "orphan" AND "disease" AND "privacy" and were restricted to those published in English.
Results: Two authors (PP, MN) independently evaluated the results for relevance to the topic, and any disagreements were resolved through discussion with all authors. The selected documents were analyzed further. We identified 5 high-risk situations in which the privacy of patients with rare diseases can be breached.
Photos: Rare diseases are poorly understood. Consequently, patient photos serve a critical role in education, are routinely displayed at scientific meetings, and are published in clinical articles. However, modern feature-recognition technology makes it possible to identify individuals shown.
Geolocation: Regulatory agencies often require data on patients from their own country to approve new interventions. When there are so few patients, publishing these data can be sufficient to identify the individuals.
Registries: Patients often participate in rare disease registries. Publishing data from these registries, taken with other patient characteristics, can possibly identify the individuals.
Reidentification: The technology available to identify individuals through cross-matching anonymized data from multiple sources can be used to identify individuals with rare disease in publications since the number of patients is so small.
Unintentional disclosure: In patient/caregiver perspectives authored by patients/caregivers, which are receiving growing interest, it is not always clear to the authors that they are voluntarily giving up their privacy.
Conclusions: Patient privacy is paramount. Guidance is needed to balance information essential to advance understanding and treatment of rare diseases while maintaining patient privacy.

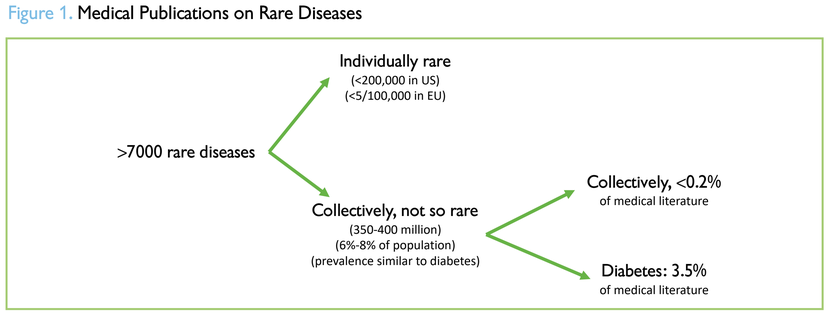
Keywords

Disclosure, Ethics, Patients, Policies, Privacy, Rare Disease

Introduction

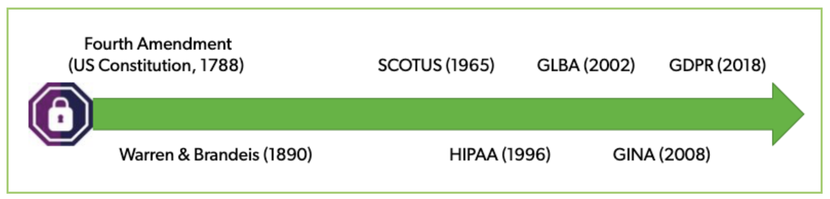
- While rare diseases individually affect only a few patients, collectively they are not rare.
  - Collectively, rare diseases affect 350-400 million people globally—6%-8% of the population (comparable to diabetes).
- Nevertheless, only 0.2% of medical literature is devoted to rare disease compared with 3.5% to diabetes **(Figure 1)**.

Figure 1. **Medical Publications on Rare Diseases**



- Patient privacy is an important factor in the conduct of clinical trials and in exchange of information.
- The concept of individual privacy in the US dates back to the Fourth Amendment of the US Constitution **(Figure 2)**.[1,2]

Figure 2. **Timeline for Privacy in the US**



GDPR, General Data Protection Regulation; GINA, Genetic Information Nondiscrimination Act; GLBA, Gramm-Leach-Bliley Act; HIPAA, Health Insurance Portability and Accountability Act; SCOTUS, Supreme Court of the United States.

- As each rare disease has very few patients, maintaining individual privacy becomes a challenge with the advancements in technology.
- These challenges are compounded in rare diseases because:
  - There is very little information on each rare disease.
  - The few patients with each rare disease are spread globally.
  - Critical information exchange needs to happen across borders.[3]
  - The risk of reidentification is high.[4]
- Although medical publication guidelines (eg, ICMJE, COPE, JAMA) have recommendations for protecting patients' rights to privacy and require consent for publication from any potentially identifiable patient, there are still many risks that are exacerbated in rare diseases.
- Despite the myriad of laws around privacy, not one specifically addresses patient/individual privacy in medical publications.

Objectives

- The goals of this project were:
  - To identify potential hurdles in maintaining individual privacy in medical publications on rare diseases.
  - To offer potential solutions to address the privacy concerns.

Methods

- A search was conducted in PubMed using the search terms "rare" OR "orphan" AND "disease" AND "privacy."
- The search was limited to the 10 years from January 1, 2010, to December 31, 2020 and restricted to those published in English and those in human patients.
- The results were supplemented by a similar search on Google.
- Two authors (PP, MN) independently evaluated the results for relevance to the topic, and any disagreements were resolved through discussion with all authors.

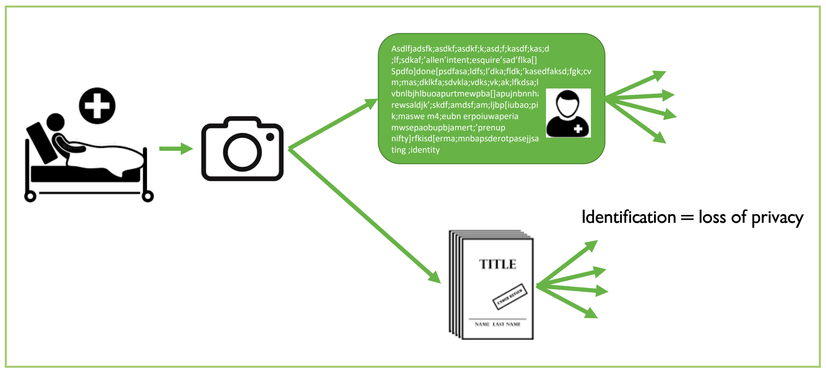
Results

- We identified 5 high-risk situations in which the privacy of patients with rare diseases could be breached.

Photos

- Patient photos serve a critical role in clinical education, especially in rare diseases where there is so little information available.
  - These photos provide clinical features and help develop natural histories in diseases.
  - The photos are also critical for understanding the impact, efficacy, and safety of interventions.
- Patient photos are, therefore, routinely displayed at scientific meetings and, subsequently, published in clinical articles.
  - These are then disseminated, without restrictions, via attendees, journal subscribers, and those who buy the articles **(Figure 3)**.
  - Thus, there is an increased risk for loss of privacy each time this information is shared.

Figure 3. **Loss of Privacy Through Medical Conferences and Literature**



Geolocation

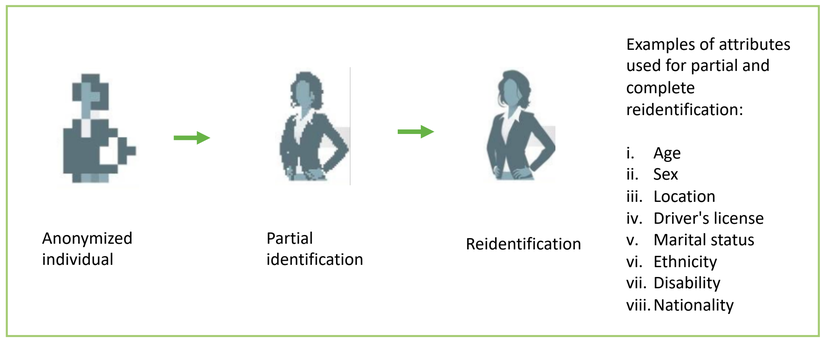
- Regulatory agencies often require data on patients from their own country to approve new interventions.
- Since there are very few patients with any particular rare disease, publishing these data can be sufficient to identify the individuals **(Figure 4)**.
- Precise, longitudinal geolocation information is impossible to anonymize and hence presents a great risk to patient anonymity.

Figure 4. **Risk of Identification Increases When Geolocation Is Well Defined**

Reidentification

- The technology available to identify individuals through cross-matching of anonymized data from multiple sources can be used to identify individuals with rare disease in publications.
- Since the number of patients is so small, it makes the reidentification easier and riskier **(Figure 5)**.

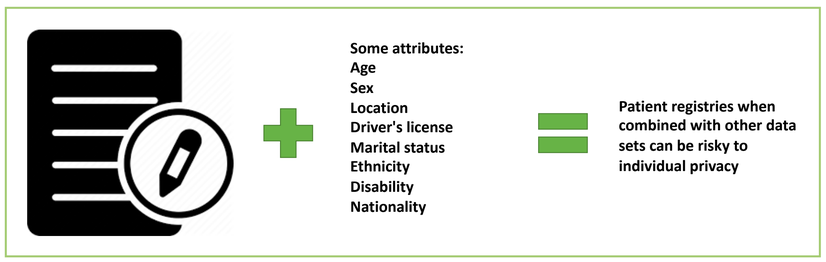
Figure 5. **Anonymized Information Can Be Reidentified**



Registries

- Patients often participate in rare disease registries.
- Publishing data from these registries, taken with other patient characteristics, can possibly identify the individuals **(Figure 6)**.

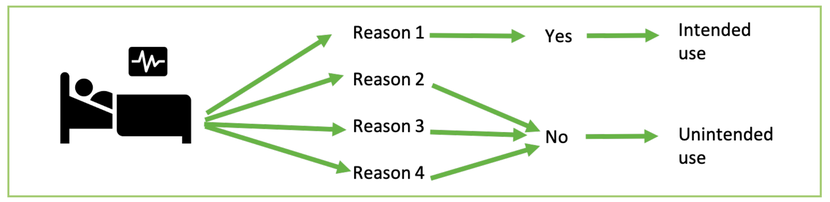
Figure 6. **Patient Registries**



Unintentional disclosure

- In patient/caregiver perspectives authored by patients/caregivers, which are receiving growing interest, it is not always clear to the authors that they are voluntarily giving up their privacy **(Figure 7)**.

Figure 7. **Unintentional Disclosure by Patients and Improper Use of Patient Information for Unspecified Purposes**



Summary

These 5 high-risk situations are summarized in **Table 1**.

Table 1. **Summary of the 5 High-Risk Situations in the Privacy of Patients With Rare Disease**

Category		Trial characteristics
1	Photos	Rare diseases are poorly understood. Consequently, patient photos serve a critical role in education, are routinely displayed at scientific meetings, and are published in clinical articles. However, modern feature-recognition technology makes it possible to identify these individuals.
2	Geolocation	Regulatory agencies often require data on patients from their own country to approve new interventions. When there are so few patients, publishing these data can be sufficient to identify the individuals.
3	Reidentification	The technology available to identify individuals through cross-matching of anonymized data from multiple sources can be used to identify individuals with rare disease described in publications since the number of patients is so small.
4	Registries	Patients often participate in rare disease registries. Publishing data from these registries, when taken with other patient characteristics, can lead to the identification of the individuals.
5	Unintentional disclosure	In patient/caregiver perspectives that are authored by patients/caregivers, it is not always clear to the authors that they are voluntarily giving up their privacy.

Limitations

- Single database of PubMed (supplemented with a Google search): It is the only widely used, freely accessible database of medical literature.
- Medical literature on PubMed is peer-reviewed.
- Search limited to articles in English: This can be addressed through original articles in English being translated and republished in other language journals.
- Small sample size of 212 articles for detailed analysis.

Discussion

- Because rare diseases collectively affect about 6%-8% of the global population, the risk and issue of individual privacy is underscored in rare diseases.
- With increased interest in patient/caregiver perspectives authored by patients/caregivers, it is important to recognize that it is not always clear to the authors that they are voluntarily giving up their privacy. A customizable omnibus consent form can be a useful tool in this regard.
- The technology available to identify individuals through cross-matching anonymized data from multiple sources can be used to identify individuals with rare disease from information in publications. Ensuring patient privacy can be handled by using software and tools that create synthetic data. There are techniques and algorithms that create data sets remarkably similar to the real data set, with 98%-99% proven accuracy.
- Regulatory agencies often require data on patients from their own country for approving new interventions. When there are so few patients, publishing these data can be sufficient to identify the individual. Using a broad area (eg, Middle East) instead of a narrower region (eg, Abu Dhabi) is a good privacy-focused alternative. Trial sponsors should discuss this privacy issue with regulatory authorities to agree on a path forward.
- Patient registries have distinctive designs and frameworks based on their purpose and use. They often contain a lot of sensitive information.
  - Transparency with the patients regarding the purpose of the registry is an important part of patient privacy.
  - Limiting metrics that are of secondary value can reduce the risk of patient information breach.
  - The fewer data you have, the fewer items you need to protect. An effective way to achieve this is to reduce the permutation and combination of micro data in data sets. It can be immensely helpful in discouraging breaches of data.

Conclusions

- Privacy is a fundamental human right enumerated in major international and regional regulations.
  - In light of rapid technological changes, it is important to revisit the current regulations for checks and balances to ensure patient privacy.
- While the authors highly value the need for more medical publications in rare diseases, we also are committed to respecting the right to privacy of patients participating in trials, those in registries, subjects of case studies, etc.
- Care must be taken to ensure privacy while moving disease understanding forward.
- Current privacy protections are inadequate and outpaced by technological advancements.
  - It is important to take a step toward changing the landscape of personal data security in rare disease medical publication.
- The medical publications industry can take a lead in developing new policies that will lead to more effective privacy regulations.

References

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