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Family ties

RAGNHILD ØRSTAVIK

ragnhild.orstavik@tidsskriftet.no
Ragnhild Ørstavik, MD, PhD, assistant editor-in-chief of the Journal of the Norwegian Medical Association and senior researcher at the Norwegian Institute of Public Health.

Case studies involving genetic disorders concern more than just the patient. Clinicians and the editors of the Journal of the Norwegian Medical Association must therefore exercise particular caution when publishing such material.

Photo: Einar Nilsen

Imagine a patient who has taken a genetic test for hereditary breast cancer. She is in her early thirties. Her mother died young from breast cancer, and she has three sisters with whom she has little contact. Most cases of this disease are hereditary. Each of the sisters therefore has a 50 % likelihood of having the same gene variant. The disease is preventable, but only if you know your own gene variant. However, the patient does not want to talk to her sisters – she would find it too taxing.

This example is adapted from an article in the American Journal of Medical Ethics and a subsequent discussion on the regulations in the United States (1). In Norway, genetic testing is subject to a statutory requirement for counselling, but it is up to the patient whether their family should be informed of possible risks (2). As a general rule, healthcare personnel cannot provide genetic information to persons other than the patient unless they have the consent of the patient (2, 3). Discussions have taken place on whether the legislation should be changed, so that regardless of consent, it should still be possible to provide such information for severe and preventable diseases (3). For the time being, the law remains unchanged. The rationale is based on the desire to protect both the right not to know and the relationship of trust between doctor and patient (3). What if the aforementioned sisters were to read a case history in this journal and realise that the patient being discussed is their sister? Put

more succinctly, the principle is the same: the authors of the article have, via the journal, passed on information to the sisters about their risk of having a genetic disease.

The Journal of the Norwegian Medical Association regularly publishes case studies where the condition turns out to be genetically determined and the inheritance is known. The severity can range from completely benign to highly complex (4, 5). Some case studies are of rare or recently discovered diseases that other doctors must be informed of (6). We in the editorial team are aware that it is difficult to anonymise a detailed case study to the point that it is guaranteed that no one – not even close family – will be able to recognise the person concerned, something that is also provided for in the declaration of consent that the patients must sign (7).

> ***«If the disease in question is hereditary, we should nevertheless be as certain as we can be that family members will not be able to recognise the patient»***

If the disease in question is hereditary, we should nevertheless be as certain as we can be that family members will not be able to recognise the patient and thus learn that he or she may be a carrier of a disease-causing gene variant. Or, of course, that people outside the family do not realise that it is not just the person or persons discussed who may be affected. This issue can be relevant for all hereditary conditions, but in practice it primarily applies to conditions with dominant inheritance. Such diseases are characterised by their multi-generational nature. In some cases, a newly discovered disease of this type will be the result of a new mutation, in which case only the patient's children are at risk.

As editors, we are very careful to anonymise case studies involving genetic diseases. All details that could potentially be used to identify a patient, and which are not strictly necessary for understanding the medical history, should be removed (7). If the text is to be supplemented with a family tree (pedigree), this should also be made as anonymous as possible – for example by using gender-neutral symbols where the line of inheritance is not linked to biological sex (8). As is standard practice, everyone mentioned in the case study must sign our consent form. This also applies if the only information that emerges is that she or he has or does not have the disease in question (8). It is rarely advisable to change factual information, even demographic data, as this can introduce errors into the research literature (8).

In the case described above, it would probably be easy to anonymise the case history, because hereditary breast cancer is relatively common. The problems are more likely to arise where rare or recently discovered conditions are concerned. However, it is precisely such cases that are important for the field of medicine to publish as a case study. The potential benefit then has to be weighed against the risk. In special cases where it is known that the immediate family are unaware of the condition, consideration can be given to finding another way to provide them with information.

REFERENCES

1. Laberge AM, Burke W. Duty to warn at-risk family members of genetic disease. Virtual Mentor 2009; 11: 656–60. [PubMed]

2. LOV-2003-12-05-100. Lov om humanmedisinsk bruk av bioteknologi m.m. (bioteknologiloven). https://lovdata.no/dokument/NL/lov/2003-12-05-100 Accessed 11.5.2023.

3. Fystro JR. Når jus og moral møtes. Tidsskr Nor Legeforen 2020; 140. doi: 10.4045/tidsskr.19.0799. [PubMed][CrossRef]

4. Drivenes JL, Grimalt R, Betz RC. Ugreelig hår. Tidsskr Nor Legeforen 2022; 142. doi: 10.4045/tidsskr.21.0888. [PubMed][CrossRef]

5. Rustad CF, Tveten K, Braathen GJ et al. En kvinne i 50-årene med langvarig muskelsvakhet. Tidsskr Nor Legeforen 2022; 142. doi: 10.4045/tidsskr.21.0038. [PubMed][CrossRef]

6. Midtvedt Ø, Stray-Pedersen A, Andersson H et al. En mann i 60-årene med kondritt og benmargssvikt. Tidsskr Nor Legeforen 2022; 142. doi: 10.4045/tidsskr.21.0370. [PubMed][CrossRef]

7. Tidsskriftet.no. Forfatterveiledning. Anonymisering og informert samtykke. https://tidsskriftet.no/annet/anonymisering-og-informert-samtykke Accessed 11.5.2023.

8. Flanagin A, Bauchner H, Fontanarosa PB. Patient and Study Participant Rights to Privacy in Journal Publication. JAMA 2020; 323: 2147–50. [PubMed][CrossRef]

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