Letter

Can the Revised "Guidelines for Genetic Tests and Diagnosis in Medical Practice" Exterminate the Spector of Genetic Exceptionalism over Medical Records?

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Genetic exceptionalism over medical records is implementing access restrictions on genetic information that are not in place for general medical information on medical records[1], which may inhibit sharing of the information “between clinical departments and between physicians and co-medicals” [2].

In March 2022, the Japanese Association of Medical Sciences (JAMS) revised the "Guidelines for Genetic Tests and Diagnosis in Medical Practice" and added a new statement that “the results of genetic testing and content of genetic counseling should likewise generally be documented in medical records, similar to other clinical data” [2]. Will this eliminate genetic exceptionalism from the medical record in Japan?

Returning to past guidelines in 2003, the Japanese societies related to genetic medicine jointly published the "Guidelines for Genetic Testing,” which stipulated that general medical and genetic information linked to a specific individual should, in principle, be stored separately[3]. Eight years later, the JAMS formulated the first edition of the "Guidelines for Genetic Tests and Diagnosis in Medical Practice" [4]. The guidelines stated that "if a genetic test is performed for diagnosis of an examinee already showing signs of symptoms, the test results should be recorded in the medical documents as any other clinical test, and should be in principle shared by all physicians and healthcare experts involved in the care of the examinee”. On the other hand, it also stated that “in case the content of genetic counseling can infringe on the privacy issue, thoughtful response is required, for example, by describing and keeping the contents of counseling separate from the usual medical record.” There was no clear description of other genetic information, such as asymptomatic carrier testing and presymptomatic testing

Until around 2011, when the first edition of the JAMS guidelines was prepared, the purpose of genetic testing was mainly limited to the diagnosis of single-gene disorders and prenatal diagnosis using chromosomes in amniotic fluid, while genetic testing with next-generation gene sequencing was not widely available[5]. In recent years, Japanese medical institutions have

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increasingly utilized genetic information, as exemplified by insurance coverage of comprehensive genomic profiling for cancer patients[5]. Simultaneously, electronic medical records have spread rapidly in Japan, and medical information is shared among many medical departments[7]. Thus, more medical professionals in more fields than before are now practicing medicine based on genetic information, and revising the JAMS guidelines is considered appropriate.

In 2020, the authors conducted an interview survey of clinical geneticists in Japan. They reported that many medical institutions implemented access restriction to genetic information on medical records either by 1) segregated storage as required in the "Guidelines for Genetic Testing" (*storage segregation*); 2) controlling specific items and entries in the electronic medical records with passwords or other means so that only authorized individuals can see them (*access control*), or both[1]. Clinical geneticists who favored access restriction cited the absence of guidelines as the need for access restriction[1].Although the revised JAMS guidelines recommend not to implement the storage segregation as a method of managing medical records and the issue of the storage segregation was explained in detail in the Q&A of the guidelines, they made no clear mention of the access control despite its prevalence in many institutions[2, 8]. Therefore, the authors expect that access restriction will continue to exist in the form of access control, leading to be a potential inhibitor of information sharing among healthcare professionals.

So, if the JAMS guidelines prohibit access control as well as storage segregation, will the problem be solved? According to our study[1], clinical geneticists expressed concerns about the possible discrimination patients would face if their genetic information was disclosed inappropriately by healthcare providers who lack sufficient knowledge of genetic information handling[1]. While efforts to eliminate the possibility of discrimination based on genetic information are important, education of healthcare professionals is also crucial and more practical, as the revised JAMS guidelines stated "all healthcare professionals who may have access to genetic information, which contains genetic characteristics in addition to other clinical information, should be educated and trained in terms of basic knowledge about genetics, confidentiality, and appropriate handling of personal genetic information" [2]. Whether genetic exceptionalism, symbolized by access restrictions on medical records, continues or disappears will depend on how appropriately the intent of the guidelines are understood and implemented.

Acknowledgement

This work was supported by JSPS KAKENHI Grant 19K24269.

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Received 15 June 2022

Final version accepted 28 July 2022