The Importance and Evolution of Informed Consent

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Informed consent has been officially around for over half a century with its first legal discussion beginning in 1905 and its establishment by law in 1957[1]. Whether this sounds young or old, the advances and spread of modern medicine and research have undoubtedly made informed consent now a universal principle of medical and research ethics.

Taking the term apart literally, “informed” means the individual has information, and “consent” refers to voluntary agreement, together meaning that the individual has the information to make an educated decision on their own[2]. The purpose of informed consent is to ensure that the individual has an understanding of the procedure, its benefits and limitations, as well as its possible

outcomes[3]. However, this principle was initially premised on surgical procedures and invasive research studies of the 20th century where physical harm was the main factor to account for.

Thus, the 20th century concept of informed consent did not sufficiently address factors that were newly introduced by genetic testing. Though the fundamental values of autonomy and nonmaleficence were there, supplemental reform was needed in order to cover other important aspects such as next steps that may be needed for an individual's health management and the impact genetic testing can have on an individual's family. With the rise of genetic testing in the early 2010s, the American College of Medical Genetics and Genomics (ACMG) recognized this gap and released recommendations on what consent for genome and exome sequencing should entail.

ACMG's points of consideration for consent include but are not limited to **the understanding of the importance of genetic counseling, secondary (or incidental) findings of high clinical**

significance that may be identified, what results the individual will or will not receive, potential benefits and risks, the limitations of testing, possible implications for family members, and distinction between testing for clinical and research use[3].

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To go over a few of these points, first, secondary findings have become an integral part of the informed consent process for genomic sequencing tests. In the case that secondary findings are opted into, it is important that the individual understands not only what secondary findings are and what secondary findings of high clinical significance mean, but also the potential for any secondary findings of initially low clinical significance to change to be of

high clinical significance with new scientific evidence in the future[4].

In addition, a potential risk of genetic testing that is often addressed is privacy and genetic discrimination for employment and insurance coverage. Hence, laws, such as the Genetic Information Non-Discrimination Act (GINA), have been established to protect individuals from such discrimination. Nevertheless, individuals are advised regarding the very modest but non-zero chance that confidentiality cannot be guaranteed[5].

Implications for family members can also be a particularly sensitive matter. Genetic test results may suggest carriers up the individual's family tree and/or have a large impact on family planning since gaining knowledge of certain hereditary factors. While the individual may feel obligated to share information with other family members, the family members may not actually want to know such information, which is usually where there can be conflict and disruption of family relationships[6].

Ultimately, the definition of “informed” can be subjective, as each individual has a different level of health literacy and a different standard for the extent of information they would like to know. This makes it important and difficult not to understate or overstate the benefits and risks of genetic testing.

Evident in how informed consent has been transformed and adapted over the years, informed consent is now moving away from an event that happens once-and-for-all and instead moving towards an ongoing collaborative process between doctor and patient or researcher and participant with regular clarification and updates6. This way, we can work towards a safe and effective application of genetic testing on the basis of honesty, transparency, and trust[7].

References

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