

NEWS

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GENOMIC MEDICINE GETS CLOSER TO THE CLINIC

The Health 2030 Genome Center (Genome Center) has obtained a major accreditation extension for human genome sequencing and its medical application. The accreditation concerns the integration of an online platform that facilitates the filtering and interpretation of genetic variants to support clinical decision-making. This new tool reduces the complexity of human genome data and brings it closer to clinical use.

Genomic medicine uses an individual's genome to understand the genetic factors behind diseases, in order to improve prevention, diagnosis and personalised treatment. The Universities and University Hospitals of Berne, Geneva and Lausanne, as well as the École Polytechnique Fédérale de Lausanne, took a major initiative in this area of medicine in 2017. They created the Genome Center, a cutting-edge genomics centre based at the Campus Biotech in Geneva capable of sequencing hundreds of human genomes per week, and a hub to promote and support genomic medicine in Switzerland.

The Genome Center obtained ISO 15189:2013 certification in May 2021, enabling the human genome, exome and transcriptome sequencing tests to be carried out on samples from hospitals and clinics. However, the interpretation of vast and rich sequencing data is complex and time-consuming, even for experienced clinicians. This is why the Genome Center has extended its accreditation to include the use of an online platform provided by a digital health company capable of fast and robust interpretation of the human genome to support clinicians in determining the genetic diagnosis.

A Genome Interpretation Tool

Developed by Congenica, the leading international company in the field, the platform acts as an initial filter for extracting genetic variants with potential clinical relevance. Thanks to this extension, clinicians can shortlist genetic variants with potential implications for patients' health, enabling them to make informed decisions about diagnosis, treatment and patient care. Technically, the digital platform compares the millions of lines of an individual's genome sequencing data with various resources to evaluate the potential pathogenicity of variants. It can also focus on specific sets of genes or pathways based on the clinical indication. Keith Harshman, Chief Operations Officer at the Genome Center, explains: This flexibility allows to adjust interpretation according to the clinician's needs. This is speeding up data processing".

A Promising Future for Genomic Medicine

Katrin Männik, Head of Genomics Strategy at the Genome Center, expresses her vision for the future of genomic medicine in Switzerland. "The use of the Congenica platform is an important step towards the democratisation of genomics in Switzerland. There is currently a huge gap between the feasibility to generate raw sequencing data and the application to the individual patient, which often discourages clinicians from ordering genetic analyses. Recent updates in the Swiss Federal Law on Human Genetic Analyses (LAGH) have broadened access to some genetic tests, allowing for example more physicians to order tests specifically to cardiology or oncology. The medical device certification of the Congenica platform will accelerate the adoption of genomic testing and facilitate the difficult work of variant interpretation for healthcare providers in their specific medical fields."



The Genome Center, as a multi-institutional player, has a not-for-profit service vocation that enables it to focus its efforts on improving healthcare based on advances in genomics research. With the expansion of its services, it is helping the Swiss clinical community to provide a more accurate interpretation of genomic information, facilitating integration of genomic applications into healthcare practices and consequently enabling more precise and personalised medicine.

About the Health 2030 Genome Center

The [Health 2030 Genome Center](#) is a national genomics competence center. Its mandate is to promote personalized health and genomic medicine by supporting and facilitating both large-scale human genomics research projects as well as the application of genomics in hospitals and clinics. The Center, located at Campus Biotech in Geneva, provides expertise and infrastructure, including ISO 15189–accredited DNA and RNA sequencing and data analysis services, to national genomics initiatives, universities, hospitals, clinics and health care providers.

Genome Center Partners:



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