

Genomics For Everyone

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Data collections using genomic or proteomic analyses often have to be produced from relatively small numbers of specimen. Particularly in case of rare diseases, the size of the accessible cohorts is frequently small. Therefore, it is very important to make research data FAIR = findable, accessible, interoperable and reusable. This was the motivation for the Center of Molecular Medicine (CeMM) and the Medical University Vienna to initiate Genom Austria (www.genomaustria.at) in 2014.

In contrast to initiatives in several other countries concentrating on medical aspects, Genom Austria's aim is to create a large, comprehensive and quality-assured Austrian genome database of volunteers, with the intention to spur public interest in genome research and to create a valuable infrastructure for high-quality research. Data management guarantees the database to be openly accessible, easily findable, compatible and reusable.

To establish the required infrastructure of the database and knowhow of next generation sequencing and data processing, 20 healthy volunteers were invited to actively participate. Their DNA was collected and subjected to whole genome sequencing. Results have been made openly accessible on the Genom Austria homepage and database. The project raised high attention and in the meantime, several hundred volunteers declared their interest. In the next 5 years, Genom Austria is planning to sequence the genomes of 1000 Austrians, and use the data to assemble an Austrian reference genome. This representation of the Austrian population on genome level will be relevant in medical applications and studies. In addition to genomic data, the database shall comprise personal (e.g. age, origin, education, socioeconomic status and personal history) and phenotypic (gender, height, weight and biometric features) information. This provides the basis for interdisciplinary research on a multitude of scientific questions. The Genom Austria database contains highly sensitive genetic and personal information. Therefore, volunteers have to sign an informed consent prior to their participation and the project is approved by the Ethics commission of the Medical University of Vienna.

Undoubtedly, genome research has the potential for major influences on society, business developments, the health care systems, etc. It is therefore important to involve citizens and decision-makers beyond the simple donation of samples. They should understand the principles, ethical considerations and consequences of genome research and actively contribute to the future applications of new technologies such as genome sequencing or genome editing. An essential prerequisite for the participation in future research developments will be an excellent education of people, providing them with clear and understandable information about where research is going and foster public discussions between experts and laypersons. This requires free access to scientific developments and ideas. With the creation of a FAIR genome and personal data collection, Genom Austria will contribute to this accomplishment.