

Genomics for Everyone

Christine Mannhalter on behalf of Genom Austria

Abstract

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**. 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.

Background on Genomics

The **Genome** comprises the complete genetic information encoded in the DNA of an organism. The human genome is organized in 23 pairs of chromosomes, of which one set is inherited from the mother and one from the father. The human genome contains roughly 20.000 protein coding genes, which are responsible for the phenotype. The basic unit of DNA are four nucleotides.

What is written in our genome?

- Ancestry and heritage
- Physical traits and appearance
- Predispositions to diseases, fitness, etc.

The genome in every individual is a unique fingerprint of a person. It is the most personal information about an individual.

Genom Austria

A scientific and cultural pioneering and educational project for genome sequencing, which was initiated in 2014 and has been funded by the **CeMM Research Center for Molecular Medicine**

The **goal** is a **quality-controlled open access research data base** including

- Genomic information
- Additional personal information (age, origin, education, socioeconomic status, medical history, biometric features)

Genom Austria data-collection is approved by the **Ethics Commission of the Medical University**

FAIR Genom Austria Database

The **Genom Austria data base** will follow the **FAIR principles** under consideration of **ethical and accessibility requirements**:

- **Findable**
Online data base on the world wide web

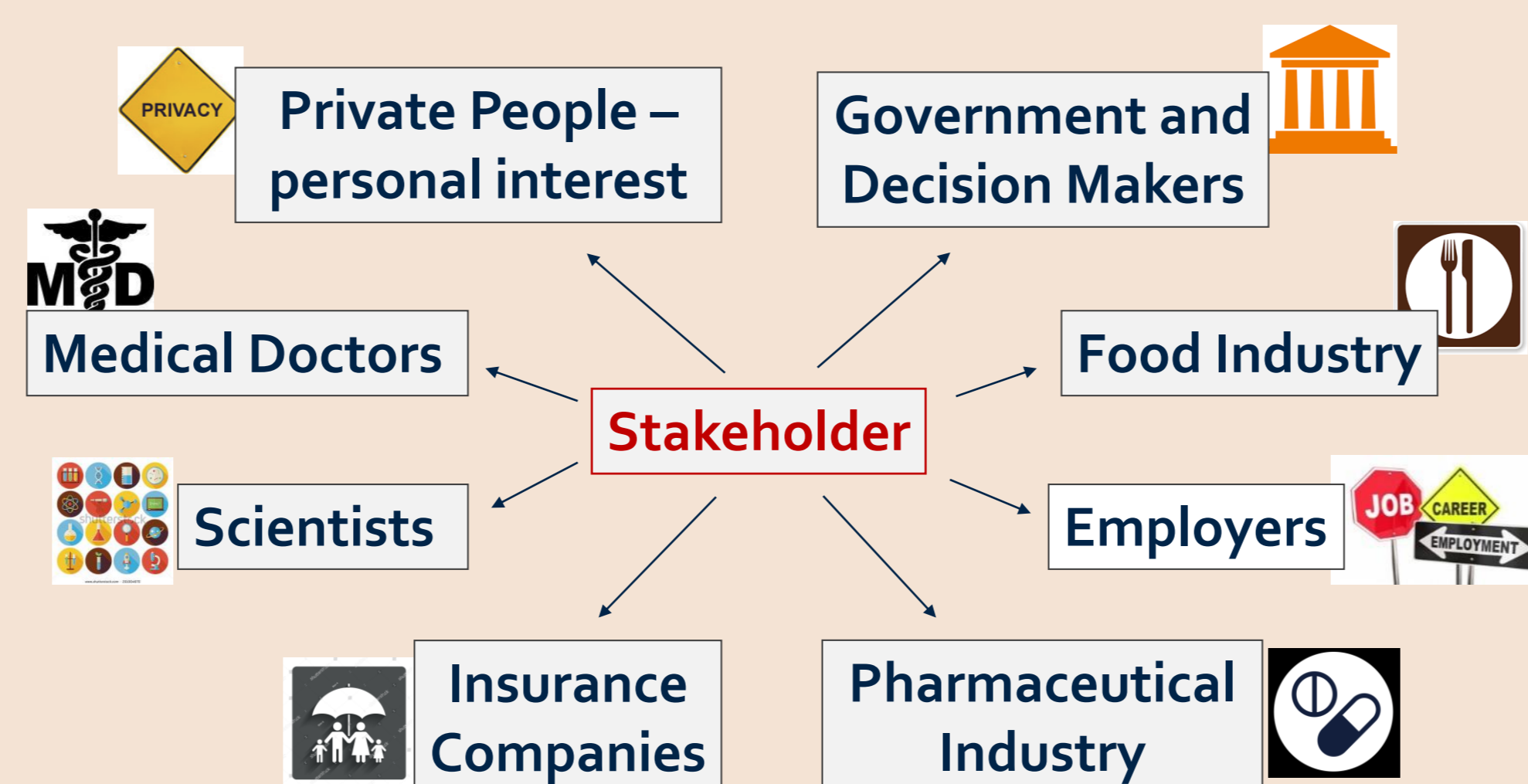
- **Accessible**
Accessible to everyone. Open access to researchers. Accessibility for industry, employers and the public sector has to fulfil certain requirements, particularly regarding data protection issues.

- **Interoperable**
Compatible and linked with already existing data collections

- **Reusable**
The data base will have the potential to grow and be used repeatedly for many projects

Stakeholder of Genomic Data

Depending the Stakeholders, certain restrictions of Open Science are necessary!



Background on Ethics and Legal Regulation

Genetic information is highly sensitive data and has to be handled accordingly. Misuse of sensitive data is an **ethical dilemma**.

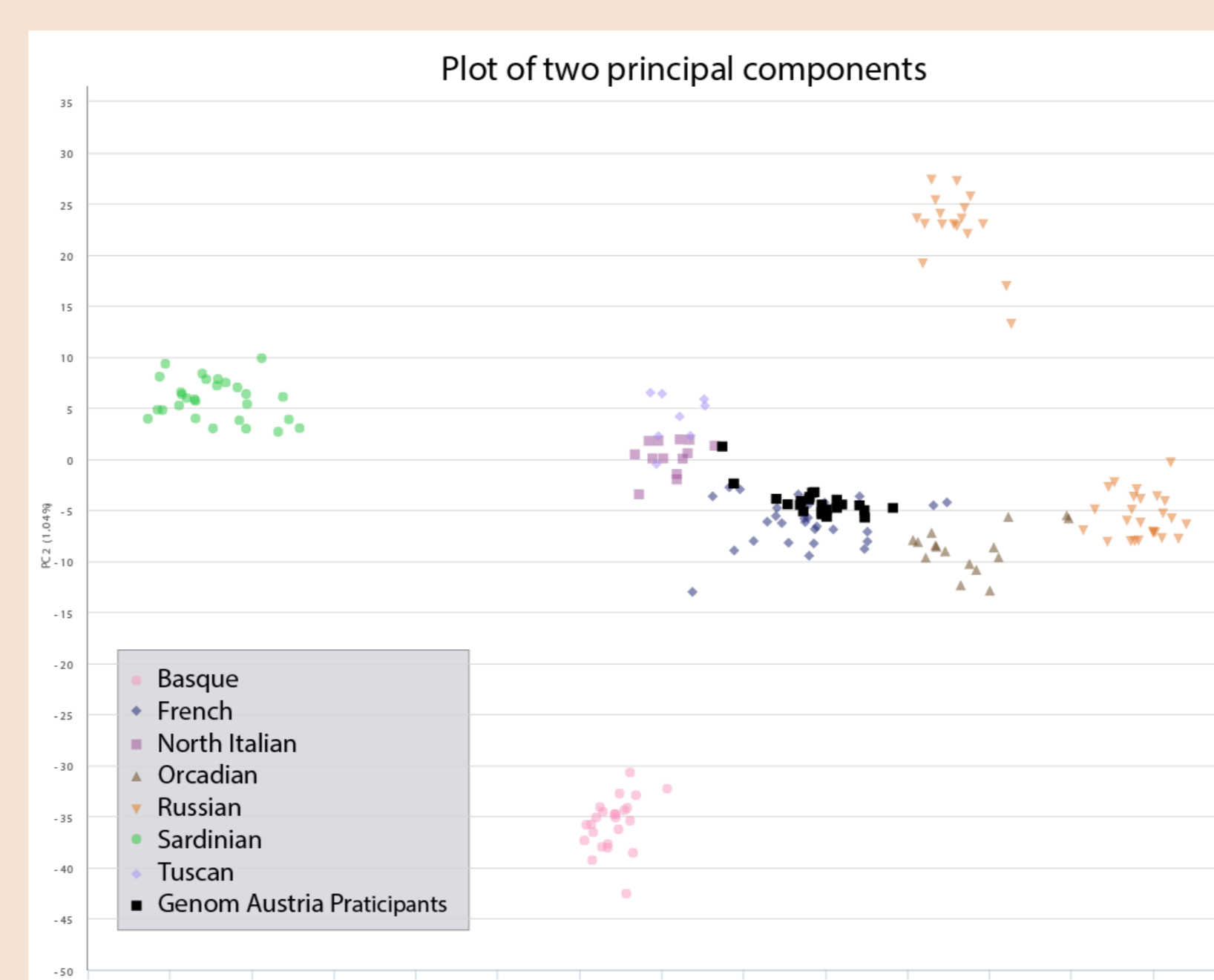
Legal regulations regarding the use of human samples and human genomic data in research and/or medicine varies between countries.

In May 2018, a **general data protection regulation (GDPR)** will go into effect. GDPR will have to be implemented in all countries of the European Union.

Genom Austria

- Genomic data linked to additional personal data are **anonymized** before they are published open access in the data base
- Access for insurances, employers, or industry will be **restricted** and **controlled**

Geographic Presentation of the Austrian Population according their genetic features



Conclusions

Open science has significant benefits

- Enables **more efficient** research and **saves costs**
- FAIR principles allow the **transparent and sustainable use** of research data

Open science has limitations

- The open science concept is not fully compatible with **highly sensitive data**, such as genomic data
- The **privacy of the person** has to be guaranteed and legal and ethical aspects have to be considered
- **Data protection regulation (GDPR)** has to be respected

Genom Austria is the first quality-controlled open access research data base built to enable interdisciplinary research

The Benefit of a Genom Austria Database

- **Illustrate the human equality and diversity**
Only 0.1% of the genomic sequence is different between two individuals
- **Build a reference genome for medical purposes**
A "matched" reference allows a more precise and personal treatment of patients
- **Connection of Natural Sciences with Humanistic and Social Studies**
Besides medicine and life sciences other disciplines show great interest in genomic data
- **Public Engagement and Education**
The public has the right to be informed of and the duty to reflect on the 21st century genomics, its applications and its consequences

Collaborators

Overall Responsibility: Christine Mannhalter

Steering Board: Christiane Druml, Markus Hengstschläger, Helga Nowotny, Michael Speicher, Giulio Superti-Furga

Project Team: Christoph Binder, Christoph Bock, Kaan Boztug, Anita Ender, Ulrich Jäger, Franco Laccone, Thomas Perkmann, Eva Schweng, Elisabeth Simböck

Advisory Board: Matthias Beck, Walter Berka, Meinrad Busslinger, Hans-Christoph Duba, Ulrike Felt, Gabriele Fischer, Barbara Horejs, Ulrich Körtnner, Markus Müller, Michael Nentwich, Peter Pakesch, Christina Peters, Barbara Prainsack, Eva Schlegel, Barbara Streicher, Johannes Zschocke

Host Institutions:

- CeMM Research Center for Molecular Medicine
- Medical University of Vienna

