

LEGAL HELPDESK

Q&A NO. 1

What are the legal provisions applicable to genetic testing and especially to whole genome sequencing as well as whole exome sequencing in the clinic in Austria?

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Question no. 1

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Subject: **Genetic testing, Gene Technology Act, Gentechnikgesetz, Informed consent**

1. Gentechnikgesetz (GTG) and genetic testing for medical purposes in Austria

COMMENT

In Austria, the subject of genetic testing for medical purposes is regulated by the **Gentechnikgesetz**, the Gene Technology Act, also known as the GTG¹, first published July 1994 (BGBl. Nr. 510/1994, came into force in 1995) and most recently altered in 2022 (through federal law BGBl. I Nr. 8/2022). The most relevant provisions of the above-mentioned GTG for the topic at hand are those contained in Section IV of the GTG (Articles 64 to 79)².

Genetic testing, according to Article 4(23) of the GTG, as translated by the European Society of Human Genetics³ (ESHG) corresponds to “[...] laboratory analysis giving evidence on concrete properties regarding number, structure or sequence of chromosomes, genes or DNA-fragments or else regarding DNA-products and specific chemical modifications thereof, thereby, due to new scientific knowledge and technical progress, providing information on a carrier status, a risk of a disease, an existing disease or a course of an illness or of a therapy on man”⁴.

The GTG explicitly prohibits interventions to the human germ line (Article 64) and authorizes human genetic testing for medical purposes in accordance with the state of the art in science and technology (Article 65) as well as genetic testing for scientific or educational purposes (Article 66). The GTG describes four types of genetic testing for **medical purposes**: genetic testing to “[...] determine an existing disease, prepare for treatment or monitor the course of treatment [...]” (authors’ translation), based on information about concrete somatic changes of number, structure, sequence or chemical modification of chromosomes, genes or DNA-segments (type 1) and to “[...] detect an existing disease that is based on a germline mutation” (type 2) (authors’ translation). Types 3 and 4 cover testing for healthy individuals who wish to determine their predisposition for a disease (in particular, the predisposition for the potential future onset of a genetically based disease) or to establish the carrier status of a disease, for which, due to new scientific knowledge and technical progress, prophylaxis or treatment either are (type 3) or are not available (type 4).

¹ *Bundesgesetz, mit dem Arbeiten mit gentechnisch veränderten Organismen, das Freisetzen und Inverkehrbringen von gentechnisch veränderten Organismen und die Anwendung von Genanalyse und Gentherapie am Menschen geregelt werden (Gentechnikgesetz – GTG)*. Available from: [RIS - Gentechnikgesetz - Bundesrecht konsolidiert, Fassung vom 11.03.2024 \(bka.gv.at\)](#) (accessed: 11/03/2024).

² For the purposes of this document, when other sources are not referenced, the GTG as well as other Austrian Law acts were translated into English unofficially using *Google Translate* and/or *DeepL Translate*.

³ Available from: [AustrianGeneTechnologyAct_English.pdf \(eshg.org\)](#) (accessed: 11/03/2024).

⁴ Preimplantation genetic diagnosis and is regulated by the Reproductive Medicine Act, i.e., the *Bundesgesetz, mit dem Regelungen über die medizinisch unterstützte Fortpflanzung getroffen werden*, known as the *Fortpflanzungsmedizingesetz* or FMedG.

In these latter two types, testing may only be carried out in facilities approved for this purpose⁵ by the Ministry of Health and only at the instigation of a specialist (medical specialist trained in human genetics or an attending or a diagnosing medical specialist), as described in Article 68.

According to Article 69(1), genetic analyses within types 2, 3 and 4 – including those carried out as part of a prenatal examination – may only be pursued after the subject has provided written confirmation that they have been informed in advance by a specialist trained in human genetics/medical genetics or a specialist responsible for the indication area⁶ about the “nature, scope and significance” of the testing and, from a detailed explanation of all test results, medical facts and possible medical, social and psychological consequences (Article 69(4)) must be able to provide a **free and informed consent** to proceed with it. The GTG also provides orientations regarding recommending relatives of the subject to pursue genetic testing (Article 70).

The sharing of test results data obtained from genetic testing follows strict rules, set in Article 71(4). These data can only be passed to staff at the facility where the data have been collected (providing they have had direct involvement with the collection, processing and evaluation of these data), the subject of the data, the legal representative of the subject of the data (according to Article 69(2)), the physician who has initiated the genetic tests and the attending physician as well as any other person whom the data subject has explicitly given a written consent to do so (with written revocation of this consent possible at any time)⁷.

In practice, authors report that there are two main avenues for patients to get genetic testing in Austria. Patients either proceed to “[...] seek treatment by doctors specialized in the disease area most relevant for them” (for example, “[...] a patient with Lupus symptoms is assigned to rheumatology, and genetic diagnosis can be advised and proceeded from there on. When test results come in, the lab sends the results back to the doctor who requested the diagnosis, e.g. the rheumatologist, who then communicates the results to the patient.”) or, alternatively, patients might choose to “[...] undergo genetic diagnosis directly via a human genetics laboratory, which then discloses the result to the patient directly”⁸. Thus, direct-to-consumer genetic testing is outlawed in Austria⁹, when carried out for medical purposes by companies which are based in Austria. Note that, in Austria, the profession of “genetic counsellor” does not exist. Professionals who carry out genetic testing patient-oriented services are medical physicians with a sub specialization in Medical Genetics (since 2007, or Medical Biology before that)¹⁰.

⁵ Listed on the *Genanalyserregister*, available here: [Genanalyserregister gemäß § 79 Abs. 1 Z 1 GTG - KVG \(verbrauchergesundheit.gv.at\)](https://www.genanalyserregister.gv.at) (accessed: 22/03/2024).

⁶ KALOKAIRINO, Louiza, et al. "Legislation of direct-to-consumer genetic testing in Europe: a fragmented regulatory landscape." *Journal of Community Genetics* 9 (2018), pp. 117-132, p. 122.

⁷ See also SATZINGER, Gabriele. "Genetische Analysen-Die Rechtslage in Österreich." *Journal für Neurologie, Neurochirurgie und Psychiatrie* 7.4 (2006), pp. 14-18.

⁸ MODELHART, Antonia, et al. "Lived experiences of genetic diagnosis for rare disease patients: a qualitative interview study." *Orphanet Journal of Rare Diseases* 19.1 (2024), p. 4.

⁹ PRAINSACK, Barbara. "The power of prediction: How personal genomics became a policy challenge." *Österreichische Zeitschrift für Politikwissenschaft* 40.4 (2011), pp. 401-415, p. 403

¹⁰ SCHWANINGER, Gunda, et al. "The genetic counseling profession in Austria: Stakeholders' perspectives." *Journal of Genetic Counseling* 30.3 (2021), pp. 861-871. Other professionals, however, may be “laboratory managers”, as explained in Article 68a.



According to Article 99 of the GTG, a *Gentechnikbuch* (Book on Gene Technology) may be published. Today, multiple chapters of this book (which constitutes soft law¹¹) are available in German¹². For the subject-matter of this question, chapters 1, 2 and 4 are of particular interest.

The Austrian Society for Human Genetics (*Österreichische Gesellschaft für Humangenetik*) provides an informed consent form for genetic testing¹³. In it, issues such as obtaining patients' consent to proceed with sample/data re-analysis to potentially detect new findings, based on technological progress of genome-wide sequencing (such as genome sequencing) are addressed. Also, the form covers sample and data repurposing for educational and scientific purposes.

2. Additional Resources

Additional resources that may be helpful.

Legal

- Council of Europe, Convention for the Protection of Human Rights and Dignity of the Human Being with regard to the Application of Biology and Medicine: Convention on Human Rights and Biomedicine, 1999. Austria is not a party to this Convention. Nevertheless, Austrian legislation is very much in line with article 12 of the Convention, which states that "Tests which are predictive of genetic diseases or which serve either to identify the subject as a carrier of a gene responsible for a disease or to detect a genetic predisposition or susceptibility to a disease may be performed only for health purposes or for scientific research linked to health purposes, and subject to appropriate genetic counselling".
- Additional Protocol to the Convention on Human Rights and Biomedicine concerning Genetic Testing for Health Purposes, Council of Europe, 2008 (CETS 203). Austria is not a signatory State to this Protocol. Nevertheless, it provides great insight into the legal and ethical issues that concern the topic at hand.

Other

- Guidelines and Opinions from the Austrian Society for Human Genetics (*Österreichische Gesellschaft für Humangenetik*), available from:
http://www.oegh.at/index.php?option=com_content&view=category&layout=blog&id=9&Itemid=16
(German only).
- Resources from the *Bundesministerium für Bildung, Wissenschaft und Forschung* (BMBWF), i.e, the Federal Ministry of Education, Science and Research, available from:
<https://www.bmbwf.gv.at/Themen/Forschung/Forschung-in-%C3%96sterreich/Services/Gentechnik.html>
(German only).

¹¹ GSCHMEIDLER, Brigitte / FLATSCHER-THOENI, Magdalena "Ethical and professional challenges of genetic counseling - the case of Austria." *Journal of Genetic Counseling* 22 (2013), pp. 741-752.

¹² Available from: [Gentechnikbuch gemäß § 99 GTG - KVG \(verbrauchergesundheit.gv.at\)](https://www.verbrauchergesundheit.gv.at) (accessed: 13/03/2024).

¹³ Available from: [fb einverstaendniserklaerung allgemein 25.11.2020.pdf \(oegh.at\)](https://www.oegh.at) (accessed: 22/03/2024).



- Resources from the *Umweltbundesamt* (Federal Environment Agency), available from: <https://www.umweltbundesamt.at/> (German only).
- Bundeskanzleramt Österreich – Bioethikkommission, *Stellungnahme der Bioethikkommission zu Gen und Genomtests im Internet*, 10.05.2010, available from: https://www.bundeskanzleramt.gv.at/dam/jcr:821b891a-4217-49e7-abb2-1effe6f3fc4a/Stellungnahme_der_Bioethikkommission_zu_Gen-_und_Genomtests_im_Internet_vom_10._Mai_2010.pdf (German only).
 - Available documents (in German only) include a Statement on somatic diagnostics of tumor tissue of the BRCA1 and BRCA2 genes and other genes and Guideline on Molecular genetic diagnostics with high-throughput germline methods, such as next-generation sequencing.
 - Other relevant document: *Stellungnahme der Bioethikkommission beim Bundeskanzleramt zum Entwurf eines Bundesgesetzes, mit dem das Fortpflanzungsmedizingesetz, das Allgemeine bürgerliche Gesetzbuch und das Gentechnikgesetz geändert werden* (Fortpflanzungsmedizinrechts-Änderungsgesetz 2015 – FMedRÄG 2015), available from: https://www.bundeskanzleramt.gv.at/dam/jcr:ecbae513-5ea7-4c76-867e-6316bff33baf/FMedRAEG_2015.pdf (German only).
- Draft WHO principles for human genome data access, use and sharing, World Health Organization, 8 April 2024, available from: [who-principles-human-genome-data-access--use--and-sharing_public-consultation_8-april.pdf](https://www.who.int/publications/m/item/who-principles-human-genome-data-access--use--and-sharing_public-consultation_8-april) (English).

Disclaimer: *this commentary aims to provide a summary of the main ethical and legal issues related to the questions put by interested stakeholders and to direct them to the relevant legal provisions that are applicable. It does not, however, preclude from reading the official sources of legislation relating to the subject matters of this document as well as those quoted by the authors and does not constitute legal advice.*