

Project Short Description

Follow-up Project: Human Genetics

Project Lead: Gregor Goetz

Project Team: Gregor Goetz, Reinhard Jeindl, Andrea Fried

Quality Assurance not relevant

Duration: Q3/2026 – Q4/2026

Language: German (no Report)

Background:

Austria does not yet have a structured, criteria-based framework for human genetic services. The AIHTA project Human Genetics 2025 provided an overview of clinical indications for human genetic tests and of evidence-based reimbursement processes (with a focus on prioritisation), and carried out two pilot assessments. This work identified countries that have introduced new reimbursement processes. In England, for example, a test directory with clear indication criteria and reimbursement rules was implemented as part of a national genomics strategy, formalising the role of health technology assessment (HTA) for new and existing tests or indications with an uncertain benefit profile. In Austria, there is interest in supporting the development of corresponding processes – particularly to establish a national test directory and ensure appropriate, evidence-based service provision.

Planned approach:

The project aims to support the further development of evidence-based service provision. To this end, at least one researcher will participate, on an ad hoc basis, in DSV working groups and processes related to genetics and contribute input from an HTA perspective. This input will draw on previous AIHTA work and, where needed, ad hoc searches. These will be limited to answering specific questions based on previous work [1–4] and, if required, brief manual web searches. Comprehensive country analyses or comparisons, as well as full assessments of individual tests, are outside the scope of the project.

In addition, the project will support a workshop of the Austrian Social Insurance, providing scientific input from an HTA perspective for the development and implementation of a national test directory in Austria. The workshop is scheduled for December 2026.

References:

- [1] Goetz, G., Jeindl, R., Yoldas, B. and Colicchia, A. (2026): Genetische Untersuchungen in Österreich. Teil A: Anwendungsgebiete, internationale Erstattungsprozesse und Priorisierungskriterien [Genetic testing in Austria. Part A: Areas of application, international reimbursement processes and prioritisation criteria]. HTA-Projektbericht 176a.
- [2] Yoldas, B., Dragosits, A. and Goetz, G. (2026): Genetische Untersuchungen in Österreich. Teil B: Carrier Screening für ausgewählte genetische Erkrankungen [Genetic testing in Austria. Part B: Carrier screening for selected genetic disorders]. HTA-Projektbericht 176b.
- [3] Colicchia, A. and Goetz, G. (2026): Genetische Untersuchungen in Österreich. Teil C: Nachweis von PIK3CA/AKT1/PTEN/ESR1-Genalterationen bei HR+/HER2– Mammakarzinom [Genetic testing in Austria. Part C: Detection of PIK3CA/AKT1/PTEN/ESR1 gene alterations in HR+/HER2– breast cancer]. HTA-Projektbericht 176c.
- [4] Jeindl, R. and Mayer-Ferbas, J. (2024): Massiv-parallele Sequenzierung – Technologien zur Hochdurchsatz-Analyse genetisch-genomischer Datensätze [Massively parallel sequencing – technologies for high-throughput analysis of genetic-genomic datasets]. AIHTA Rapid Review Nr.: 015.