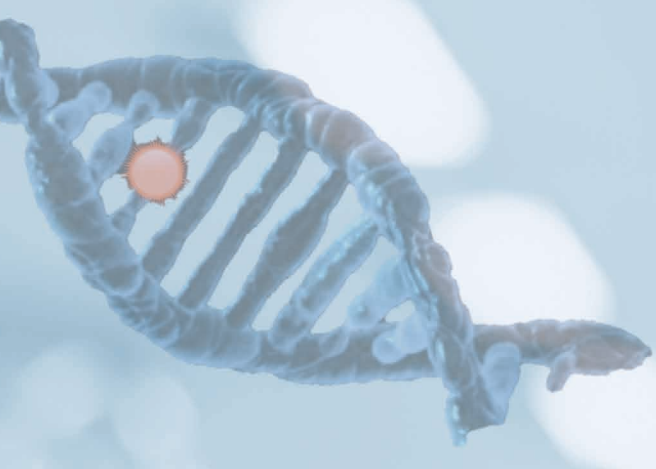
TECHNISCHE HOCHSCHULE DEGGENDORF



*The best form of therapy*

*quickly identified.*

# DIGITALIZATION IN MOLECULAR DIAGNOSTICS

AUTOMATION AND STANDARDIZATION OF MOLECULAR PATHOLOGY DATA ANALYSIS AND REPORT GENERATION IN THE FIELD OF CANCER DIAGNOSTICS.

Weitere Infos

# Abstract


**Projekttitel/ Project title:**

**Next Generation Genomic Data Analysis for Clinics & Research**

***GEDA***

**Einleitung/ Introduction:**

With a global incidence of 19.3 million and nearly 10 million related deaths in 2020, cancer is the second leading cause of death in the world. Knowledge of genes and associated mutations involved in cancer development - combined with the ability to perform cost-effective gene sequencing via Next Generation Sequencing (*NGS*) methods and bioinformatics analysis - is a powerful tool for cancer patient screening and therapy discovery. Therefore, automated and standardized bioinformatics analysis processes are urgently needed to evaluate the increasingly large number of variants and attempt to prioritize those that may be important for disease risk and progression.

**Ziel/ Aim:**

The aims of this project were the establishment of a cooperative collaboration with the Institute of Pathology in Deggendorf, obtaining an ethics vote regarding the planned research project to enable data exchange between the THD and the Institute of Pathology and the development of a basis for automated *NGS* data analysis as in-house solution for molecular diagnostics and research.

**Methode/ Method:**

Tumor sequencing today has an immense impact on the development and progress of precise and personalized treatment approaches. The most useful aspects of tumor sequencing are the identification of therapeutic targets and the discovery of additional tumor biomarkers for molecular diagnostics. After the establishment of a cooperative collaboration and successful request of an ethics vote for data sharing with the Institute of Pathology, the individual databases containing essential information for molecular pathological diagnosis with regard to detected tumor-associated variants, gene fusions and numerical chromosomal changes (Copy Number Variations) were cataloged. The most time-consuming steps currently performed manually were elicited and potential automation options were sought. Based on the *NGS* data analysis results - SNPs (Single Nucleotide Polymorphisms), InDels (Insertions and Deletions), Gene Fusions and CNVs (Copy Number Variations) - a software prototype could be developed.

**Ergebnis/ Result:**

**Projektbeteiligte/ Project participants:**

The software prototype *MolPatInGer* (MOLecular PAThologist´s INvestigator GERmany) was developed in close cooperation with the Institute of Pathology in Deggendorf to adapt the software to the needs of diagnostic laboratories. This software links the tumor-relevant information from various databases to the via *NGS* detected variants (SNPs, InDels), gene fusions and CNVs gained from NGS data, so that the time required for the search with regard to the clinical relevance, the functional relevance of the altered genes, the available therapeutic options based on the detected changes, etc. could be minimized (14-33% time savings in this sub-process). Further development of the prototype will be the subject of further projects between the THD and the Institute of Pathology.

**Projektpartner/ Project partners:**

Institute for Pathology, Molecular Pathology and Cytology (Deggendorf; Dr. Alexandra Hamberger and Dr. med. Mohren)



**Gefördert durch/ Funded by:**

**Deggendorf Institute of Technology**

## Logos/ Logos:

**Bilderreferenzen:**
https://pixabay.com/
BillionPhotos.com/stock.adobe.com
https://www.cancer.net