TRANSFERRING CODE TO DIAGNOSIS

Ce GaT
Center for Genomics and Transcriptomics

TUMOR-DIAGNOSTICS

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SUMMARY

- Molecular profiling of 551 cancer-relevant genes
- Complete sample-to-diagnosis pipeline
- Reliable detection of variants down to 20% tumor content
- Only 100 ng of starting material required
- Medical report with clinical interpretation as treatment decision support
- Valuable data for clinical studies

BACKGROUND

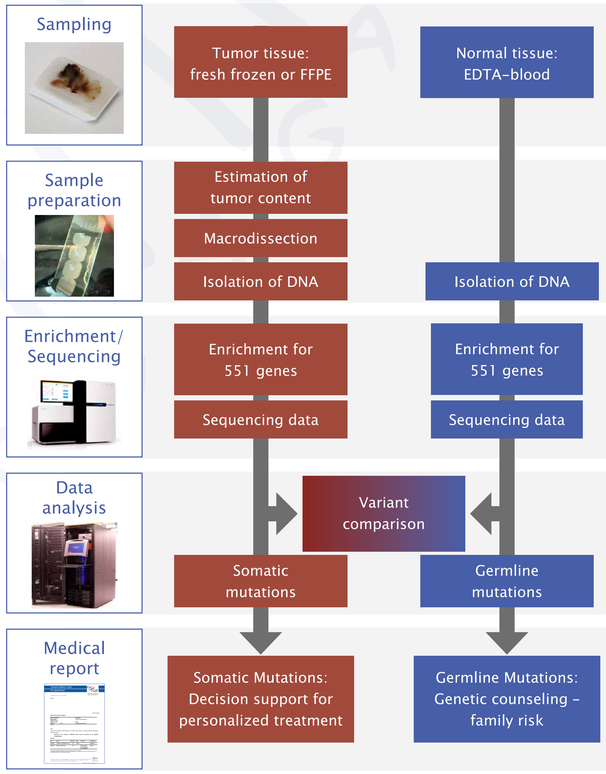
An increasing number of targeted anti-cancer drugs are becoming available, raising the requirement for comprehensive molecular profiling of tumors.

Next-generation high-throughput sequencing (NGS) offers the opportunity to analyze a huge number of genes from a small quantity of starting material.

We have developed a complete clinical diagnostic pipeline for the analysis of 551 cancer-relevant genes using next-generation sequencing.

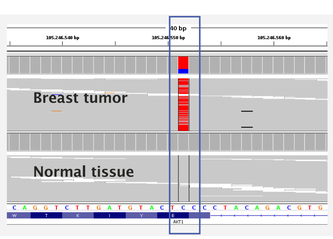
Comprehensive molecular profiling using panel-based next-generation sequencing can identify therapeutic targets in tumor patients.

WORKFLOW

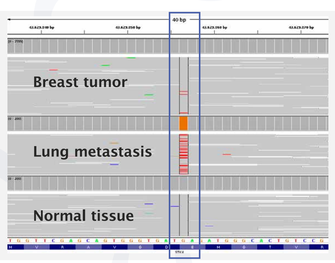


SAMPLE CASE

- 46 year old patient with locally recurrent breast cancer and lung metastasis histologically defined as adenocarcinoma of unknown primary (possibly stomach).
- Panel-based sequencing of a lung biopsy and a local breast cancer metastasis.



Somatic activating mutation E17K in *AKT1*. The mutation is present in the tumor, but not in the normal tissue (blood).



A nonsense mutation in STK4 is present in a small (but detectable) subclone of the breast tumor (Allele frequency 3 %). The same mutation is more frequent in the lung metastasis (Allele frequency 29 %).

RESULTS

- Lung metastasis is clonally related to the recurrent breast cancer (same origin).
- Activating *AKT1* mutation conferring resistance to tamoxifen
- *TP53* mutation, studies show lower benefit from anthracycline-based therapies

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