



**MIRACUM  
DIFUTURE**

Medizininformatik für Forschung und Versorgung

# **Erweiterungen von cBioPortal für das molekulare Tumorboard und Einbindung in die IT-Gesamtumgebung des Universitätsklinikums Erlangen**

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**02.07.2024**

**MIRACUM-DIFUTURE-Kolloquium**



GEFÖRDERT VOM



Bundesministerium  
für Bildung  
und Forschung

# Erweiterungen von cBioPortal für das molekulare Tumorboard





# Hintergrund: Was ist cBioPortal?

- Cancer Genomics Daten Integration: cBioPortal integriert Krebsgenom-Daten, einschließlich CNV, mRNA-Expression, Mutationen und klinische Daten aus mehreren Quellen.
- Query and Analyse Werkzeuge: Benutzer können eine Reihe von Analysen durchführen, wie z.B. die Identifizierung genetischer Veränderungen bei bestimmten Krebsarten, das gleichzeitige Auftreten und die wechselseitige Ausschließlichkeit von Genomveränderungen sowie Überlebensanalysen basierend auf Genomdaten.
- Custom Daten Upload: Eigene Daten können in die Plattform geladen werden um so deren Werkzeuge für die Visualisierung und Analyse der Daten verwenden.
- Flexibles Deployment: Alle cBioPortal Komponenten können also containerisierte Anwendungen bspw. unter Verwendung von Docker deployed werden.
- Open Source und Community-Driven Development: cBioPortal ist ein Open Source Projekt, ursprünglich entwickelt durch das Memorial Sloan Kettering Cancer Center. Durch die aktiven Maintainer und die große Nutzer-Community wird cBioPortal kontinuierlich mit Updates und neuen Funktionen versorgt.

Cerami et al. The cBio Cancer Genomics Portal: An Open Platform for Exploring Multidimensional Cancer Genomics Data. *Cancer Discovery*. May 2012; 2; 401. [PubMed](#).

Gao et al. Integrative analysis of complex cancer genomics and clinical profiles using the cBioPortal. *Sci. Signal.* 6, pl1 (2013). [PubMed](#).

de Bruijn et al. Analysis and Visualization of Longitudinal Genomic and Clinical Data from the AACR Project GENIE Biopharma Collaborative in cBioPortal. *Cancer Res* (2023). [PubMed](#).

## TMB and Immunotherapy (MSK, Nat Genet 2019)

Genomic and survival data from 1661 tumor-normal pairs from 1661 patients with various cancer types sequenced with the MSK-IMPACT assay. [PubMed](#)

Click gene symbols below or enter here



Summary

Clinical Data

Plots Beta!

Selected: 1,661 patients | 1,661 samples



Custom Selection ▾

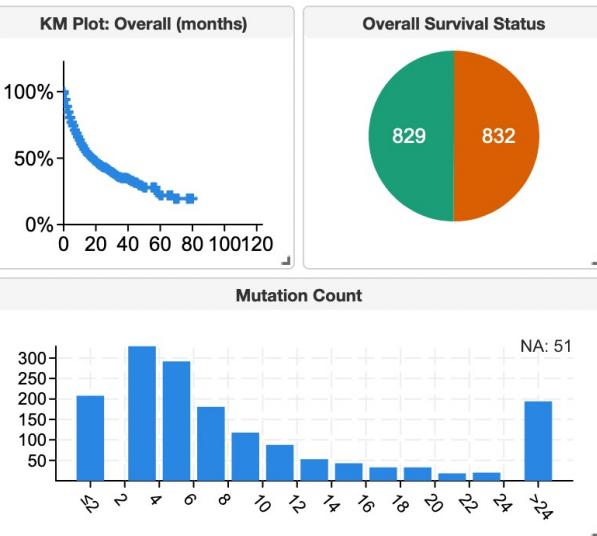
Charts ▾

Groups ▾

Cancer Type		#	Freq ▾
Non-Small Cell Lung Cancer	<input type="checkbox"/>	350	21.1%
Melanoma	<input type="checkbox"/>	320	19.3%
Bladder Cancer	<input type="checkbox"/>	215	12.9%
Renal Cell Carcinoma	<input type="checkbox"/>	151	9.1%
Head and Neck Cancer	<input type="checkbox"/>	139	8.4%
Esophagogastric Cancer	<input type="checkbox"/>	126	7.6%
Glioma	<input type="checkbox"/>	117	7.0%
Colorectal Cancer	<input type="checkbox"/>	110	6.6%
Cancer of Unknown Primary	<input type="checkbox"/>	88	5.3%
Breast Cancer	<input type="checkbox"/>	44	2.6%
Skin Cancer, Non-Melanoma	<input type="checkbox"/>	1	<0.1%
Search...		Search...	

Cancer Type Detailed		#	Freq ▾
Lung Adenocarcinoma	<input type="checkbox"/>	271	16.3%
Cutaneous Melanoma	<input type="checkbox"/>	187	11.3%
Bladder Urothelial Carcinoma	<input type="checkbox"/>	147	8.9%
Renal Clear Cell Carcinoma	<input type="checkbox"/>	121	7.3%
Colon Adenocarcinoma	<input type="checkbox"/>	85	5.1%
Glioblastoma Multiforme	<input type="checkbox"/>	82	4.9%
Upper Tract Urothelial Carcinoma	<input type="checkbox"/>	47	2.8%
Lung Squamous Cell Carcinoma	<input type="checkbox"/>	45	2.7%
Melanoma of Unknown Primary	<input type="checkbox"/>	44	2.6%
Esophageal Adenocarcinoma	<input type="checkbox"/>	39	2.3%
Oropharynx Squamous Cell Carci...	<input type="checkbox"/>	37	2.2%
Search...		Search...	

Genomic Profile Sample Counts		#	Freq ▾
Molecular Profile	<input type="checkbox"/>	1,661	100.0%
Structural variants	<input type="checkbox"/>	1,661	100.0%



Mutated Genes (1661 profiled samples)			
Gene	# Mut	#	Freq ▾
TP53	834	<input type="checkbox"/>	738 44.4%
TERT	572	<input type="checkbox"/>	519 31.2%
KMT2D	356	<input type="checkbox"/>	236 14.2%
KRAS	232	<input type="checkbox"/>	226 13.6%
PIK3CA	220	<input type="checkbox"/>	200 12.0%
ARID1A	252	<input type="checkbox"/>	190 11.4%
NF1	241	<input type="checkbox"/>	183 11.0%
PTPRT	244	<input type="checkbox"/>	176 10.6%
PREX2	52	<input type="checkbox"/>	42 10.1%
KMT2B	56	<input type="checkbox"/>	42 10.1%
BRAF	181	<input type="checkbox"/>	165 9.9%
Search...		Search...	

Structural Variant Genes (1661 profiled samples)			
Gene	# SV	#	Freq ▾
EGFR	30	<input type="checkbox"/>	29 1.7%
BRAF	10	<input type="checkbox"/>	10 0.6%
ROS1	9	<input type="checkbox"/>	9 0.5%
APC	8	<input type="checkbox"/>	8 0.5%
TP53	7	<input type="checkbox"/>	7 0.4%
FGFR3	7	<input type="checkbox"/>	7 0.4%
RB1	6	<input type="checkbox"/>	6 0.4%
TACC3	6	<input type="checkbox"/>	6 0.4%
ALK	6	<input type="checkbox"/>	6 0.4%
EWSR1	6	<input type="checkbox"/>	5 0.3%
SMARCA4	5	<input type="checkbox"/>	5 0.3%
Search...		Search...	



Metastatic Site		#	Freq ▾
NA	<input type="checkbox"/>	1	<0.1%

Oncotree Code		#	Freq ▾
NA	<input type="checkbox"/>	1	<0.1%





Patient: P-0000004, Female, Breast Cancer (Breast Invasive Ductal Carcinoma), DECEASED (3 months)

Samples: 1 P-0000004-T01-IM3, Primary (Breast)

MSK MetTropism (MSK, C)

&lt; &lt; 1 of 25775 patients &gt;

Summary

Pathways

Clinical Data



#### 4 Mutations (page 1 of 1)

  Columns ▾

Gene	Protein Change	Annotation	Mutation Type	Allele Freq	Copy #	Cohort
AKT1	E17K	● ● ● 🔥	Missense	0.55	Diploid	2%
TP53	A138Cfs*27	●	FS del	0.22	Diploid	48%
SPEN	I3661F	○	Missense	0.15	Diploid	4%
KMT2C	M812I	○	Missense	0.12	Diploid	6%

Showing 1-4 of 4 Mutations

#### 0 Structural Variants (page 1 of 1)

  Columns ▾

Gene 1	Gene 2	Status	Annotation	Variant Class	Event Info	Connection Type
There are no results.						

#### 3 Copy Number Alterations (page 1 of 1)

  Columns ▾

# Hintergrund: MIRACUM Use Case 3

## Ziele und Ergebnisse



- Ziel: Verbesserung der personalisierten Medizin durch IT-Lösungen für die Unterstützung Molekularer Tumorboards
- Umfassende Anforderungsanalyse und Mockups für eine MTB-Plattform basierend auf cBioPortal durch Buechner et al.
- Iterative Umsetzung und Evaluation → MTB-cBioPortal (ehemals MIRACUM-cBioPortal)
- Entwicklung weiterer Werkzeuge
  - MIRACUM Pipeline
  - CBPManager
  - ...
- Rollout der entwickelten Lösungen in einer Reihe von Kliniken

Buechner P, Hinderer M, Unberath P, et al. Requirements Analysis and Specification for a Molecular Tumor Board Platform Based on cBioPortal. *Diagnostics (Basel)*. 2020;10(2):93. Published 2020 Feb 10. doi:10.3390/diagnostics10020093



# MTB-cBioPortal

cBioPortal FOR CANCER GENOMICS Data Sets Web API R/MATLAB Tutorials/Webinars FAQ News Visualize Your Data About

Patient: Testpatient, Breast Carcinoma, LIVING (12 months), Recurred/Progressed (4 months)  
Samples: ① Testpatient\_01, MSI-H ②, TMB-H ③ ② Testpatient\_02, MSI-H ④, TMB-H ⑤

Summary Pathways Clinical Data MTB ClinicalTrialsGov

breast x  
BRAF x  
Recruiting x  
Germany x  
Select age...  
All  
Select patient location...  
 Set maximum distance in km  
Search

Status	Matching Criteria	Study Title	Conditions	Interventions	Eligibility Criteria	Locations
Recruiting	<ul style="list-style-type: none"> <li>Age is matching</li> <li>Gender is matching</li> <li>Found keywords: BRAF</li> </ul>	An Open-label Phase II Multicenter Study of Vemurafenib (Zelboraf®) Plus Cobimetinib (Cotellic®) After Radiosurgery in Patients With Active BRAF-V600-mutant Melanoma Brain Metastases	<ul style="list-style-type: none"> <li>Malignant Melanoma Stage IV</li> <li>BRAF V600 Mutation</li> <li>Brain Metastases</li> </ul>	<ul style="list-style-type: none"> <li>Vemurafenib</li> <li>Cobimetinib</li> </ul>	Show ▾	<ul style="list-style-type: none"> <li>Dresden: Technische Universität Dresden: undefined</li> <li>Heidelberg: Ruprecht-Karls-University of Heidelberg, Faculty of Medicine: undefined</li> <li>Tübingen: Eberhard Karls University of Tübingen, University Medical Center: undefined</li> </ul>
Recruiting	<ul style="list-style-type: none"> <li>Age is matching</li> <li>Gender is matching</li> <li>Found keywords: BRAF</li> </ul>	Enhancing Radioiodine Incorporation Into Radio Iodine Refractory Thyroid	Metastatic Thyroid Cancer	<ul style="list-style-type: none"> <li>Trametinib 2 MG [Mekinst]</li> <li>Trametinib 2 MG</li> </ul>	Show ▾	<ul style="list-style-type: none"> <li>Essen: Manuel M. Weber: Northrhine-Westphalia</li> </ul>

cBioPortal FOR CANCER GENOMICS Data Sets Web API R/MATLAB Tutorials/Webinars FAQ News Visualize Your Data About

Patient: Testpatient, Mary Jane, Female, 60 years old, Breast Carcinoma, LIVING (12 months), Recurred/Progressed (4 months)  
Samples: ① Testpatient\_01, Primary, MSI-H ②, TMB-H ③ ② Testpatient\_02, Metastasis, MSI-H ④, TMB-H ⑤

Summary Pathways Clinical Data MTB Follow-up ClinicalTrialsGov

## MTB Sessions

+ Add MTB Save Data

MTB Info

Date: 21/02/2024

State: Partial

Genetic Counseling  
 Rebiopsy

Comments

Select considered samples...

+ Delete MTB

### Therapy Recommendations

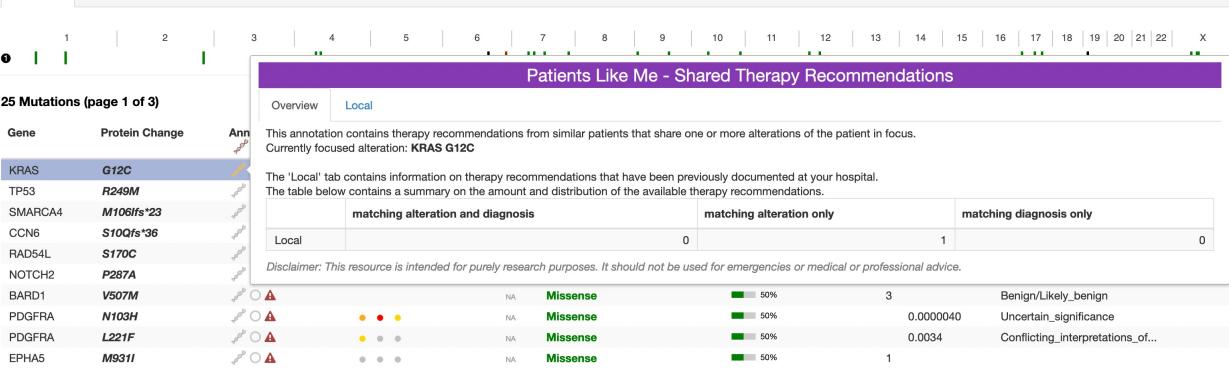
+ Add	+ Add from OncoKB	+ Add from template	Prio	Reasoning	Therapy / Trials	Comment	Evidence Level	References	Edit
				Genomic alterations: KRAS G12C ① In samples:	Clinical data / molecular diagnostics: AZD5363		Level m1C ①		<input type="checkbox"/> Edit <input type="checkbox"/> Delete

Showing 1-1 of 1

cBioPortal FOR CANCER GENOMICS Data Sets Web API R/MATLAB Tutorials/Webinars FAQ News Visualize Your Data About

Patient: H38009-19, Female, Female, 53 years old, LIVING (7 months)  
Samples: ① H38009-19

Summary Pathways Clinical Data MTB Follow-up ClinicalTrialsGov



Evaluation Study

1 of 2 patients > >

Q

dbSNP

rs121913530

rs587782329

rs372964061

rs1222128776

rs139913632

rs1415093400

rs1222128776

rs139913632

rs1415093400

### 2 Copy Number Alterations (page 1 of 1)

cBioPortal FOR CANCER GENOMICS Data Sets Web API R/MATLAB Tutorials/Webinars FAQ News Visualize Your Data About

Patient: Testpatient, Mary Jane, Female, 60 years old, Breast Carcinoma, LIVING (12 months), Recurred/Progressed (4 months)  
Samples: ① Testpatient\_01, Primary, MSI-H ②, TMB-H ③ ② Testpatient\_02, Metastasis, MSI-H ④, TMB-H ⑤

Summary Pathways Clinical Data MTB Follow-up ClinicalTrialsGov

### Follow-up data

+ Add FollowUp Save Data

### Follow-up

Therapy Recommendation

+ Add + Add from OncoKB + Add from template

Reasoning

Genomic alterations:  
KRAS G12C ①  
In samples:  
testcomment

Comment

Evidence Level

References

21/02/2024

Realized  
 Side effect

Tumor response criteria

Month	PD	SD	PR	CR
3	<input type="checkbox"/>	<input checked="" type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
6	<input type="checkbox"/>	<input type="checkbox"/>	<input checked="" type="checkbox"/>	<input type="checkbox"/>
12	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input checked="" type="checkbox"/>

Showing 1-1 of 1

Testpatient

1 of 1 patients > >

Testpatient

Q

Level m1C ①

References

Showing 1-1 of 1

Showing 1-1 of 1

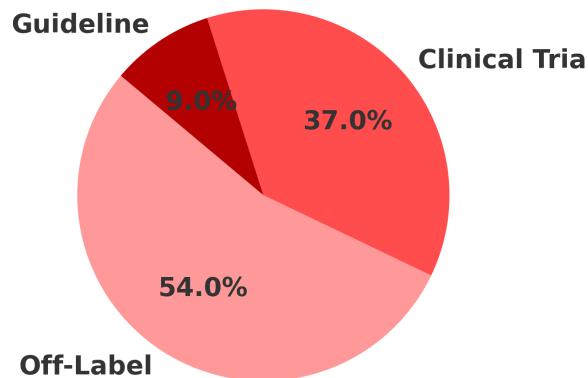


- Semi automatisierte Suche nach passenden klinischen Studien basierend auf der clinicaltrials.gov API
- Unterstützung von Dokumentation für MTB-Sitzungen, Therapieempfehlungen und dazugehörige Follow-Up Daten
- Integration von Autorisierungs Features für Dokumentations Erweiterungen
- Annotation von Alterationen mit zuvor dokumentierten Therapieempfehlungen auf der Basis ähnlicher Patienten
- Containerisiertes Deployment der gesamten Infrastruktur



- MTB-Empfehlungen beinhalten häufig den Einschluss in klinische Studien  
→ Derzeit muss händisch nach passenden klinischen Studien gesucht werden

## Therapieempfehlungen



## Unterstützung bei der Suche nach klinischen Studien

- Integration in cBioPortal
- Nutzerzentrierte Entwicklung
- Nutzung der ClinicalTrials.gov API anstelle manueller Suche

Hoefflin R et al. Transitioning the Molecular Tumor Board from Proof of Concept to Clinical Routine: A German Single-Center Analysis. Cancers (Basel). 2021 Mar 8;13(5):1151. doi: 10.3390/cancers13051151. PMID: 33800365; PMCID: PMC7962829.

Unberath P, Mahlmeister L, Reimer N, Busch H, Boerries M\*, Christoph J\*. Searching of Clinical Trials made easier in cBioPortal using Patients' Genetic and Clinical Profiles. Appl Clin Inform. 2022 Mar;13(2):363-369. doi: 10.1055/s-0042-1743560. Epub 2022 Mar 30. PMID: 35354211; PMCID: PMC8967483.


Patient: **Testpatient**, Breast Carcinoma, **LIVING** (12 months), **Recurrent/Progressed** (4 months)

Samples: **1** Testpatient\_01, MSI-H , TMB-H   **2** Testpatient\_02, MSI-H , TMB-H 

Testpatient

[Summary](#)   [Pathways](#)   [Clinical Data](#)   [MTB](#)
[ClinicalTrialsGov](#)

breast

|

BRAF

|

Recruiting

|

Germany

|

Select age...

All

Select patient location...

 Set maximum distance in km

[Search](#)

Status	Matching Criteria	Study Title	Conditions	Interventions	Eligibility Criteria	Locations
Recruiting	<ul style="list-style-type: none"> <li>Age is matching</li> <li>Gender is matching</li> <li>Found keywords: BRAF</li> </ul>	<a href="#">An Open-label Phase II Multicenter Study of Vemurafenib (Zelboraf®) Plus Cobimetinib (Cotellic®) After Radiosurgery in Patients With Active BRAF-V600-mutant Melanoma Brain Metastases</a>	<ul style="list-style-type: none"> <li>Malignant Melanoma Stage IV</li> <li>BRAF V600 Mutation</li> <li>Brain Metastases</li> </ul>	<ul style="list-style-type: none"> <li>Vemurafenib</li> <li>Cobimetinib</li> </ul>	Show: <input type="checkbox"/>	<ul style="list-style-type: none"> <li>Dresden: Technische Universität Dresden: undefined</li> <li>Heidelberg: Ruprecht-Karls-University of Heidelberg, Faculty of Medicine: undefined</li> <li>Tuebingen: Eberhard Karls University of Tübingen, University Medical Center: undefined</li> </ul>
Recruiting	<ul style="list-style-type: none"> <li>Age is matching</li> <li>Gender is matching</li> <li>Found keywords: BRAF</li> </ul>	<a href="#">Enhancing Radioiodine Incorporation Into Radio Iodine Refractory Thyroid Cancers With MAPK Inhibition: A Single Center</a>	<ul style="list-style-type: none"> <li>Metastatic Thyroid Cancer</li> </ul>	<ul style="list-style-type: none"> <li>Trametinib 2 MG [Mekinist]</li> <li>Trametinib 2 MG [Mekinist] and Dabrafenib 75 MG (2-0-</li> </ul>	Show: <input type="checkbox"/>	<ul style="list-style-type: none"> <li>Essen: Manuel M. Weber: Northrhine-Westphalia</li> </ul>



Patient: Testpatient, Mary Jane, Female, 60 years old, Breast Carcinoma, LIVING (12 months), Recurred/Progressed (4 months)

Samples: 1 Testpatient\_01, Primary, MSI-H, TMB-H 2 Testpatient\_02, Metastasis, MSI-H, TMB-H

Testpatient

Summary Pathways Clinical Data MTB Follow-up ClinicalTrialsGov

Search clinical trials

35 results have been found. The results are based on: [Search parameters](#)

Mutations: KRAS  
 Tumor Entities: Breast Carcinoma  
 Recruiting Status: Recruiting, Not yet recruiting  
 Countries:  
 Patient Age: 60  
 Patient Location: Erlangen | Bavaria | Germany  
 Max Distance From Location: 200

## Clinical Trial Search

Columns ▾



Status	Matching Criteria	Study Title	Conditions	Interventions	Eligibility Criteria	Locations
Recruiting	Age is matching Gender is matching Condition is matching Found keywords: KRAS	<a href="#">Adjuvant CAPECITABINE in High Risk PSEUDOMYXOMA PERITONEI Patients Treated With CYTOREDUCTIVE SURGERY (CRS) and HYPERTERMIC INTRAPERITONEAL CHEMOTHERAPY (HIPEC)</a>	Pseudomyxoma Peritonei	Capecitabine	<a href="#">show</a>	Milano   Fondazione IRCCS Istituto Nazionale dei Tumori di Milano   MI
Not yet recruiting	Age is matching Gender is matching Condition is matching Found keywords: KRAS	<a href="#">A Multicenter, Open-label, Phase Ib Study to Evaluate the Safety, Tolerability, Pharmacokinetics and Preliminary Efficacy of CYH33 in Combination With Endocrine Therapy With or Without Palbociclib in Patients With PIK3CA Mutant, HR+, HER- Advanced Breast Cancer</a>	Advanced Breast Cancer	CYH33 Fulvestrant Letrozole Palbociclib	<a href="#">show</a>	
Recruiting	Age is matching Gender is matching Distance to Bad Berka is 146 km Found keywords: KRAS	<a href="#">A Randomized Phase 3 Study of MRTX849 Versus Docetaxel in Patients With Previously Treated Non-Small Cell Lung Cancer With KRAS G12C Mutation</a>	Metastatic Non Small Cell Lung Cancer Advanced Non Small Cell Lung Cancer	MRTX849 Docetaxel	<a href="#">show</a>	Duarte   Research Site   California Long Beach   Research Site   California Santa Rosa   Research Site   California Whittier   Research Site   California Grand Junction   Research Site   Colorado
Recruiting	Age is matching Gender is matching Distance to Esslingen is 156 km Found keywords: KRAS	<a href="#">A Phase 2 Trial of MRTX849 Monotherapy and in Combination With Pembrolizumab in Patients With Advanced Non-Small Cell Lung Cancer With KRAS G12C Mutation</a>	Advanced Non-Small Cell Lung Cancer Metastatic Non-Small Cell Lung Cancer	MRTX849 Monotherapy MRTX849 in Combination with Pembrolizumab MRTX849 in Combination with Pembrolizumab	<a href="#">show</a>	Prescott Valley   Research Site   Arizona Tucson   Research Site   Arizona Springdale   Research Site   Arkansas Anaheim   Research Site   California San Francisco   Research Site   California
Recruiting	Age is matching Gender is matching Distance to Esslingen is 156 km Found keywords: KRAS	<a href="#">A Phase 2, Open-Label, Multicenter Study of the Combination of RMC-4630 and Sotorasib for Non-Small Cell Lung Cancer Subjects With KRASG12C Mutation After Failure of Prior Standard Therapies</a>	Non-Small Cell Lung Cancer	RMC-4630 Sotorasib	<a href="#">show</a>	Fort Myers   Florida Cancer Specialists   Florida Plantation   BRCR Medical Center Inc.   Florida Saint Augustine   Cancer Specialists of North Florida   Florida Evergreen Park   GenHarp Clinical Solutions   Illinois Baton Rouge   Hematology Oncology Clinic   Louisiana
Recruiting	Age is matching Gender is matching Distance to Esslingen is 156 km Found keywords: KRAS	<a href="#">A Phase II/III Multicenter Study Evaluating the Efficacy and Safety of Multiple Targeted Therapies as Treatments for Patients With Advanced or Metastatic Non-Small Cell Lung Cancer (NSCLC) Harboring Actionable Somatic Mutations Detected in Blood (B-FAST: Blood-First Assay Screening Trial)</a>	Non-Small Cell Lung Cancer	Alectinib Atezolizumab Pemetrexed Cisplatin Carboplatin	<a href="#">show</a>	La Jolla   University of California San Diego   California Sacramento   UC Davis; Comprehensive Cancer Center   California Denver   Rocky Mountain Cancer Center   Colorado Norwich   Eastern Connecticut Hematology and Oncology Associates; (ECHO)   Connecticut Fort Myers   SCRI Florida Cancer Specialists South   Florida



# MTB-cBioPortal

MTB Dokumentation and Verwendung standardisierter Datensätze

- MTB Dokumentation besteht derzeit häufig aus semi-strukturiertem Freitext → Verringerung der Nutzbarkeit der Daten für automatisierte Verarbeitung und strukturierte Suche

The screenshot shows a complex web-based form for a molecular tumor board. It includes fields for patient history, molecular analysis results, treatment history, potential therapies, and clinical questions. A sidebar on the right lists participants from various departments like Pathology, Radiology, and Hematology.

- Dokumentations Funktion basierend auf dem MII Erweiterungsmodul - Molekulargenetischer Befundbericht
- MTB Report mit Therapieempfehlungen, und für die Empfehlung relevante klinische Daten and Alterationen
- Zusätzliche Erweiterung für die Dokumentation von Follow-Up Daten auf der Basis einzelner Therapieempfehlungen

Renner C, Reimer N, Christoph J, et al. Extending cBioPortal for Therapy Recommendation Documentation in Molecular Tumor Boards: Development and Usability Study. *JMIR Med Inform*. 2023;11:e50017. Published 2023 Dec 11. doi:10.2196/50017



Patient: [Testpatient](#), Mary Jane, Female, 60 years old, Breast Carcinoma, **LIVING** (12 months), **Recurrent/Progressed** (4 months)

Samples: [1 Testpatient\\_01](#), Primary, MSI-H , TMB-H [2 Testpatient\\_02](#), Metastasis, MSI-H , TMB-H

[Summary](#)
[Pathways](#)
[Clinical Data](#)
[MTB](#)
[Follow-up](#)
[ClinicalTrialsGov](#)

## MTB Sessions

[+ Add MTB](#)
[Save Data](#)


### MTB Info



Date: [21 / 02 / 2024](#)

State: [Partial](#)

Genetic Counseling

Rebiopsy

Comments

Select considered samples...

[Delete MTB](#)

### Therapy Recommendations

[+ Add](#)
[+ Add from OncoKB](#)
[+ Add from template](#)

Prio	Reasoning	Therapy / Trials	Comment
	Genomic alterations: <b>KRAS G12C</b> In samples:	Clinical data / molecular diagnostics: Cancer Type: Breast Carcinoma Disease Free (Months): 4	 <b>AZD5363</b>

Showing 1-1 of 1

Showing 1-1 of 1



Patient: Testpatient, Mary Jane, Female, 60 years old, Breast Carcinoma, LIVING (12 months), Recurred/Progressed (4 months)

Samples: ① Testpatient\_01, Primary, MSI-H ⓘ, TMB-H ⓘ ② Testpatient\_02, Metastasis, MSI-H ⓘ, TMB-H ⓘ

[Summary](#) [Pathways](#) [Clinical Data](#) [MTB](#) [Follow-up](#) [ClinicalTrialsGov](#)

## Follow-up data

[+ Add FollowUp](#) [Save Data](#)

### Follow-up

### Therapy Recommendation

11.08.2022 [Edit](#)

Realized  
 Side effect

Tumor response criteria

Months	PD	SD	PR	CR
3	<input checked="" type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
6	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
12	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

Comments

[Delete](#)[+ Add](#)[+ Add from OncoKB](#)[+ Add from template](#)

#### Reasoning

Genomic alterations: KRAS G12C ⓘ  
Clinical data / molecular diagnostics: Analysis Method: Oncomine

In samples:  
①

#### Comment

Recommendation imported from OncoKB.

#### Evidence Level

Level m2A ⓘ

#### References

[34096690] Sotorasib for Lung Cancers

Showing 1-1 of 1

Showing 1-1 of 1



# MTB-cBioPortal

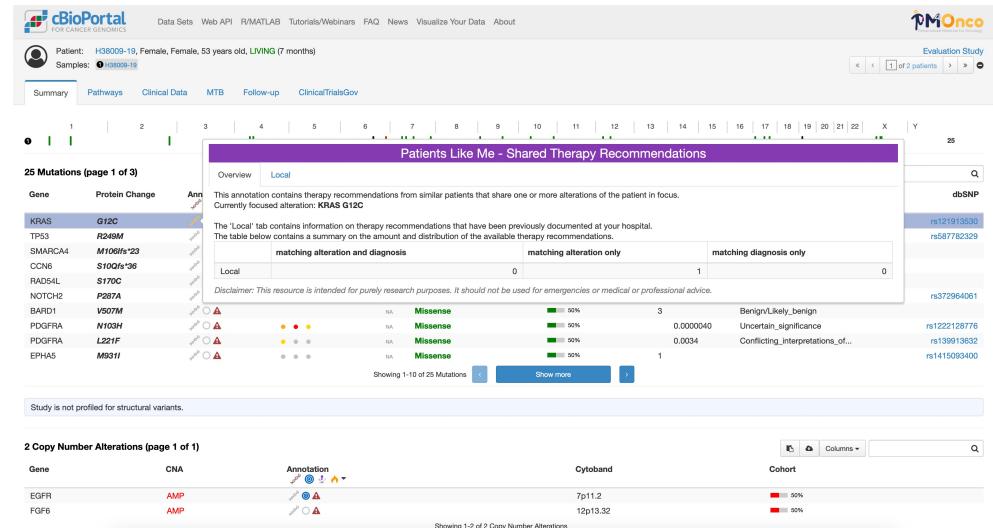
Additional annotations and reuse of therapy recommendations



- Fall Vorbereitung für MTB Sitzungen ist sehr zeitaufwändig
- Große Menge an repetitiver Arbeit durch händische Literatur- und Fall-Suche



- Entscheidungsunterstützung durch Annotation mit zusätzlichen Informationen
- Wiederverwendung von (lokal verfügbaren) Therapieempfehlungen und Follow-Up Daten



Patient: H38009-19, Female, Female, 53 years old, LIVING (7 months)  
 Samples: 1 H38009-19

[Evaluation](#)

&lt; &lt; [ 1 of 2 patients ] &gt;

[Summary](#) [Pathways](#) [Clinical Data](#) [MTB](#) [Follow-up](#) [ClinicalTrialsGov](#)


## 25 Mutations (page 1 of 3)

Gene	Protein Change	Annotation	Alteration	Cancer Type	Treatment	Evidence Level	Therapy Response	Clinical Data	Comment	Patient
KRAS	G12C		KRAS G12C	Adeno-CUP	AZD8186	m1A	3 months: NA 6 months: NA 12 months: NA	Diagnosis: Adeno-CUP Age (Years): 41 ICD-O3-Morphologie Code: 8140/3		<a href="#">support_study_2023 : H38009-23</a>
TP53	R249M									
SMARCA4	M106Ifs*23									
CCN6	S10Qfs*36									
RAD54L	S170C									
NOTCH2	P287A									
BARD1	V507M									
PDGFRA	N103H									
PDGFRA	L221F									
EPHA5	M931I									

Disclaimer: This resource is intended for purely research purposes. It should not be used for emergencies or medical or professional advice.

Showing 1-10 of 25 Mutations

[Show more](#)
[>](#)

Study is not profiled for structural variants.

## 2 Copy Number Alterations (page 1 of 1)

Gene	CNA	Annotation	Cytoband	Cohort
EGFR	AMP		7p11.2	
FGF6	AMP		12p13.32	

Showing 1-2 of 2 Copy Number Alterations

# MTB-cBioPortal

## Other Addons/Tools & Team Collaboration



KRAS G12C

Oncogenic Gain-of-function

KRAS, a GTPase which functions as an upstream regulator of the MAPK pathway, is frequently mutated in various cancer types including lung, colorectal and pancreatic cancers.

The KRAS G12C mutation is known to be oncogenic.

Biological Effect Therapeutic Implications Diagnostic Implications

Level	Alteration(s)	Drug(s)	Level-associated cancer type(s)
R1	Oncogenic Mutations	Cetuximab €	Colorectal Cancer
R1	Oncogenic Mutations	Panitumumab €	Colorectal Cancer
	Sotorasib is not authorized in the EU. More info on cancerdrugs	€	Non-Small Cell Lung Cancer
2	Oncogenic Mutations	Cobimetinib €	Erdheim-Chester Disease
2	Oncogenic Mutations	Cobimetinib €	Langerhans Cell Histiocytosis
2	Oncogenic	Cobimetinib €	Rosai-Dorfman Disease

The information above is intended for research purposes only and should not be used as a substitute for professional diagnosis and treatment.

Levels of Evidence

Onc@KB Feedback

## EU Drug Approval Status



## Data Management Tool

Ustjanzew A, Desuki A, Ritzel C, Dolezilek AC, Wagner DC, Christoph J, Unberath P, Kindler T, Faber J, Marini F, Panholzer T, Paret C. cbpManager: a web application to streamline the integration of clinical and genomic data in cBioPortal to support the Molecular Tumor Board. BMC Med Inform Decis Mak. 2021 Dec 20;21(1):358. doi: 10.1186/s12911-021-01719-z. PMID: 34930224; PMCID: PMC8686377.

# Fortführung der Entwicklung in PM4Onco

AP5



- Frühe Phase des  Projekts
- Aktuelle Arbeiten:
- Anforderungserhebung – die zuvor identifizierten Anforderungen wieder mit den Wünschen und Bedürfnissen der MTB-TeilnehmerInnen in Einklang zu bringen.
- Anforderungserhebung – Abfrage des aktuellen Stands von Implementierungs and Datenintegrations-Arbeiten für Software für die Unterstützung molekularer Tumorboards
- Evaluation – Vergleich mit den Evaluations Ergebnissen der Summary Evaluation die am Ende von MIRACUM use case 3 durchgeführt wurde
- Scoping Review – Überblick über bestehende Visualisierungs-Lösungen und Arbeiten im Tumor Board Bereich
- Und vieles mehr...

Boehm D, Strantz C, Christoph J, Busch H, Ganslandt T, Unberath P. Data Visualization Support for Tumor Boards and Clinical Oncology: Protocol for a Scoping Review. *JMIR Res Protoc.* 2024;13:e53627. Published 2024 Mar 5. doi:10.2196/53627

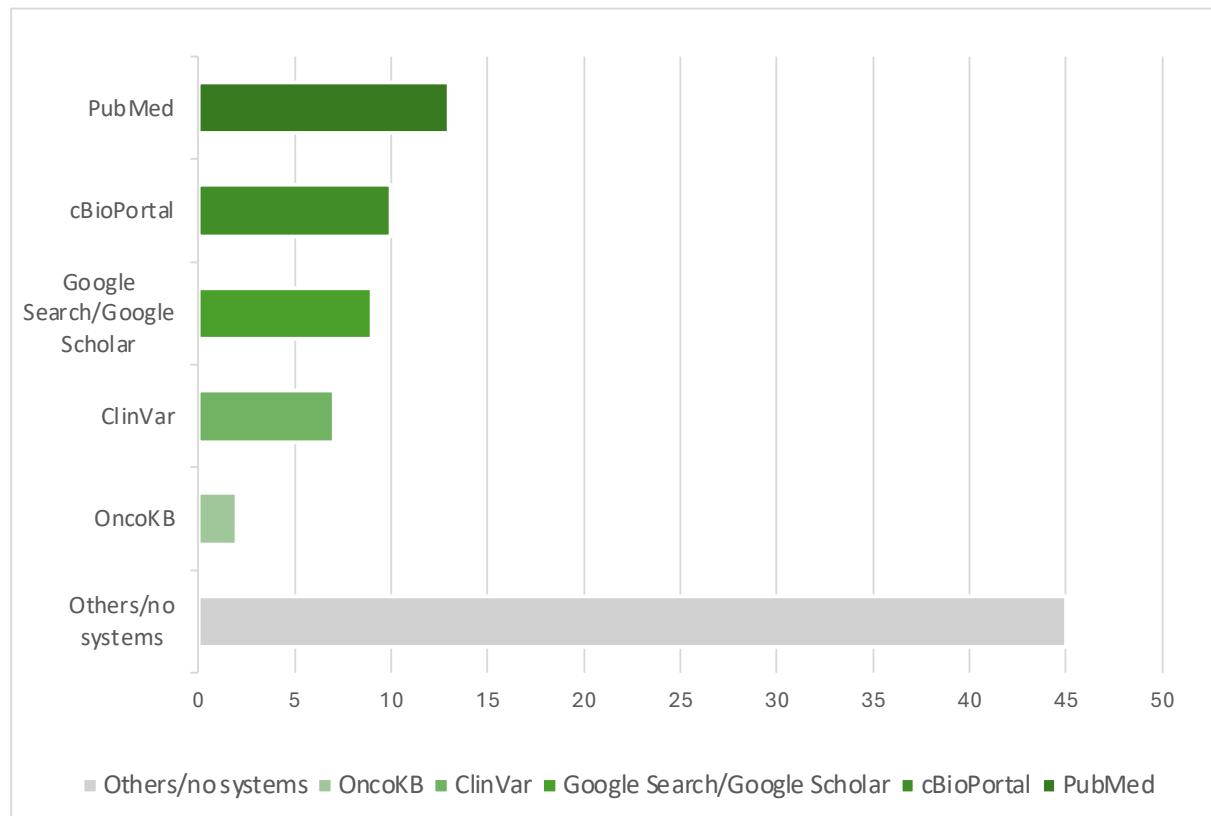
# Literatur Review und Anforderungserhebung



## Anforderungserhebung Ergebnisse – MTB Vorbereitung

- Obwohl einige Systeme häufiger als andere für die Vorbereitung verwendet werden ist die Bandbreite sehr groß
- Nutzer sind generell zufrieden mit ihren aktuellen Vorbereitungsprozessen
- Wünschen sich aber bessere Integration der genutzten Werkzeuge und weniger manuelle Suche, um die Effizienz der Vorbereitung zu erhöhen

Distribution of applied systems / applications / websites for the preparation of the MTB



# Literatur Review and Anforderungserhebung

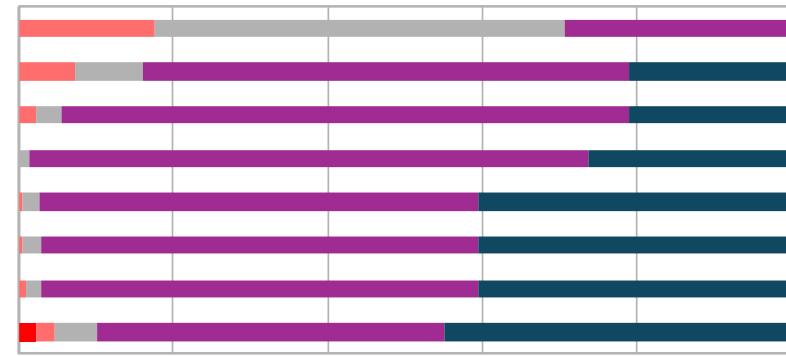


## Anforderungserhebung Ergebnisse – zusätzliche Visualisierungs-Funktionen

- Nutzer sehen einen hohen Bedarf an neuartigen Visualisierungsmethoden für fast alle Datentypen im MTB Kontext
- PROMs haben bisher eine untergeordnete Rolle in der Vorbereitung von MTBs gespielt, was die Diskrepanz zu den anderen Datentypen erklären könnte

Requirement for novel visualization methods along different data types:

- ... for PROMs standard/reference population.
- ... for PROMs single patient.
- ... for historical data.
- ... for (novel) complex biomarkers.
- ... for gene expression.
- ... for CNA/CNV.
- ... for genomic variants.
- ... for clinical data.



■ very little need ■ little need ■ partly ■ high demand ■ very high demand



# Aktuelle Entwicklungen

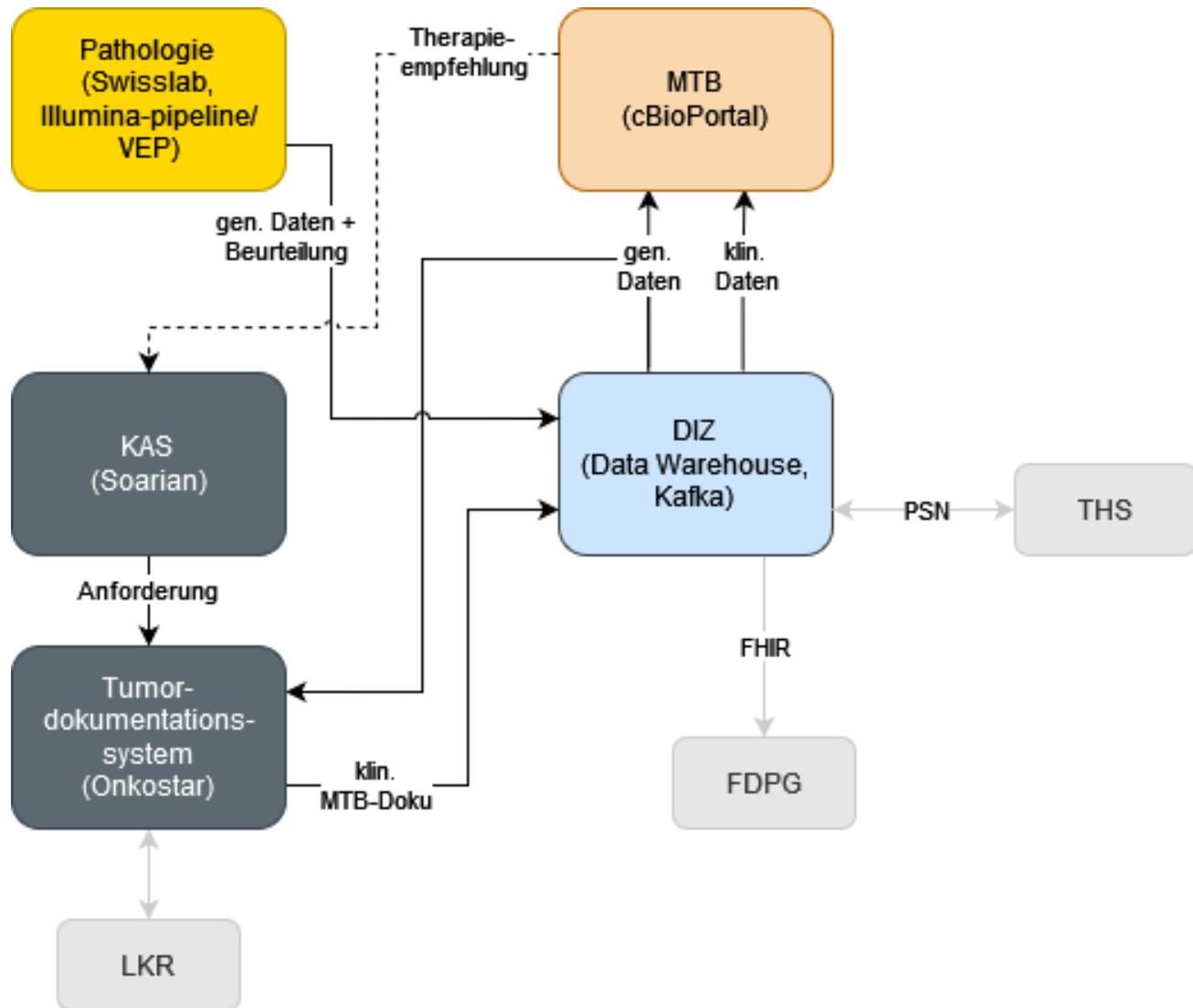
- Iterative Verbesserung der Nutzbarkeit für die Vorbereitung und Durchführung molekularer Tumorboards
  - Timeline
  - Dokumentationsfunktion
- Präsentationsfunktion
  - Aktuell laufende Masterarbeit
- Zukünftig: Integration neuer Visualisierungsfunktionen im Rahmen von PM4Onco
  - AP3 PROMs
  - AP5 Patientenähnlichkeit
  - ...

# Einbindung in die IT-Gesamtumgebung des Universitätsklinikums Erlangen





# Einbindung in die IT-Landschaft - Überblick



## Pathologie

Laborsystem: Swisslab

Panel: TSO-500

## Zentrale IT

KAS: Soarian

Tumordokumentation: Onkostar

## MTB

cBioPortal

PowerPoint

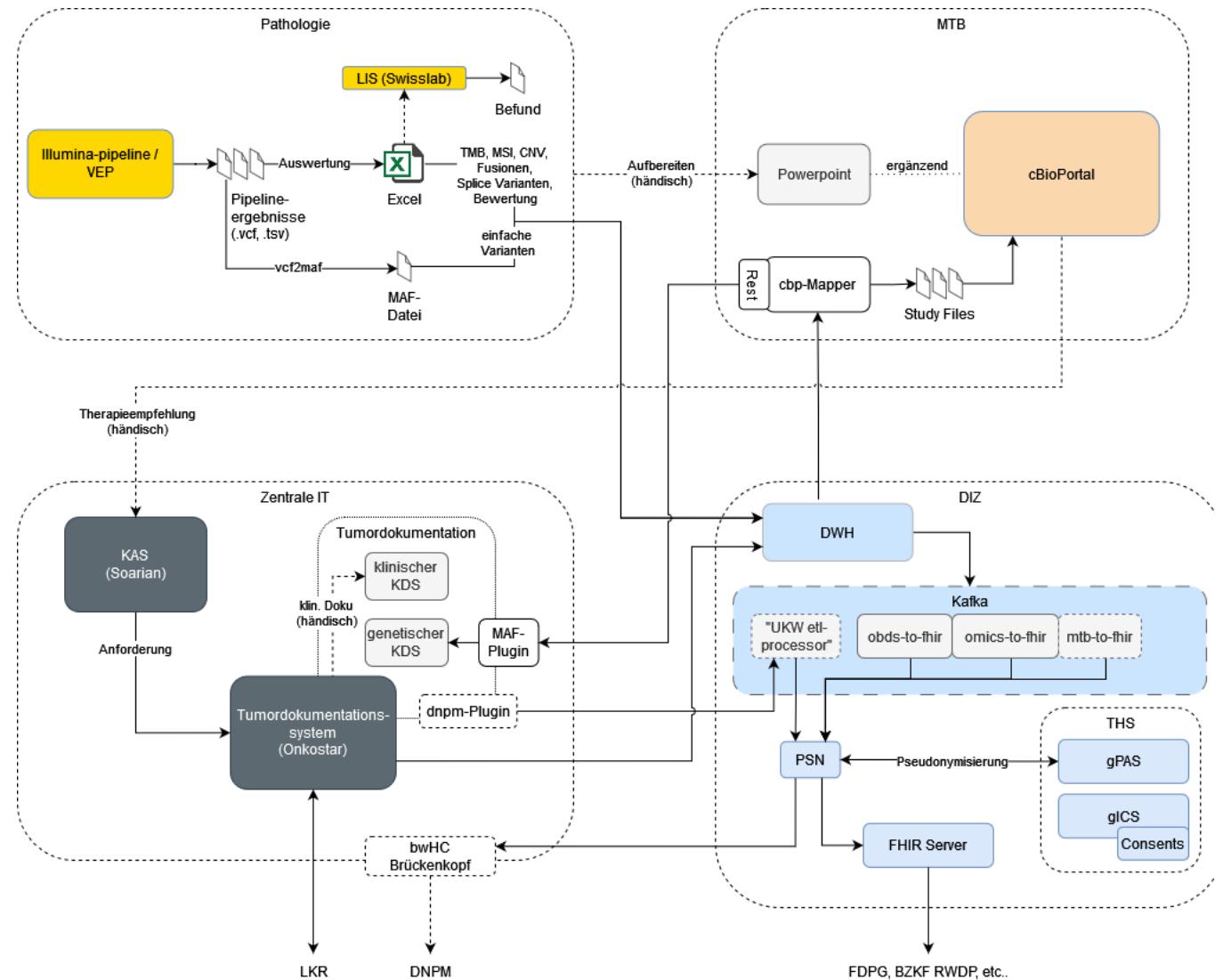
## DIZ

Data Warehouse

Kafka (ETL)



# Einbindung in die IT-Landschaft - Detailansicht



## Pathologie

Laborsystem: Swisslab

Panel: TSO-500

## Zentrale IT

KAS: Soarian

Tumordokumentation: Onkostar

## MTB

cBioPortal

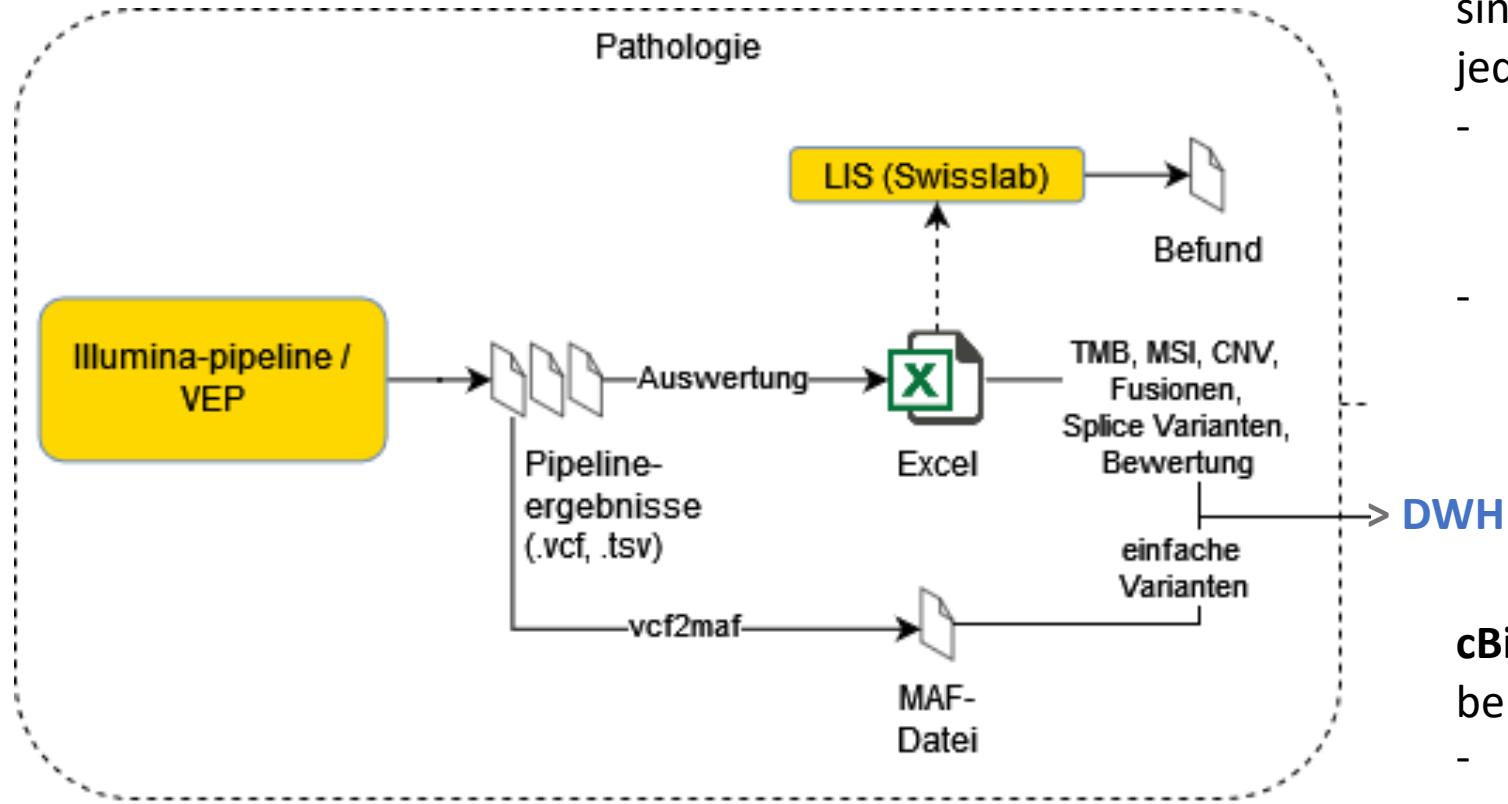
PowerPoint

## DIZ

Data Warehouse

Kafka (ETL)

# Einbindung in die IT-Landschaft - Detailansicht Pathologie



## Pipeline Ergebnisse

sind sehr umfassend; vor-annotiert, jedoch nicht bewertet

- Auswertung & Einschränkung auf relevante Mutationen durch Pathologie in „Auswertungs-Excel“
- Hinzufügen der Pathogenitätseinschätzung

## cBioPortal

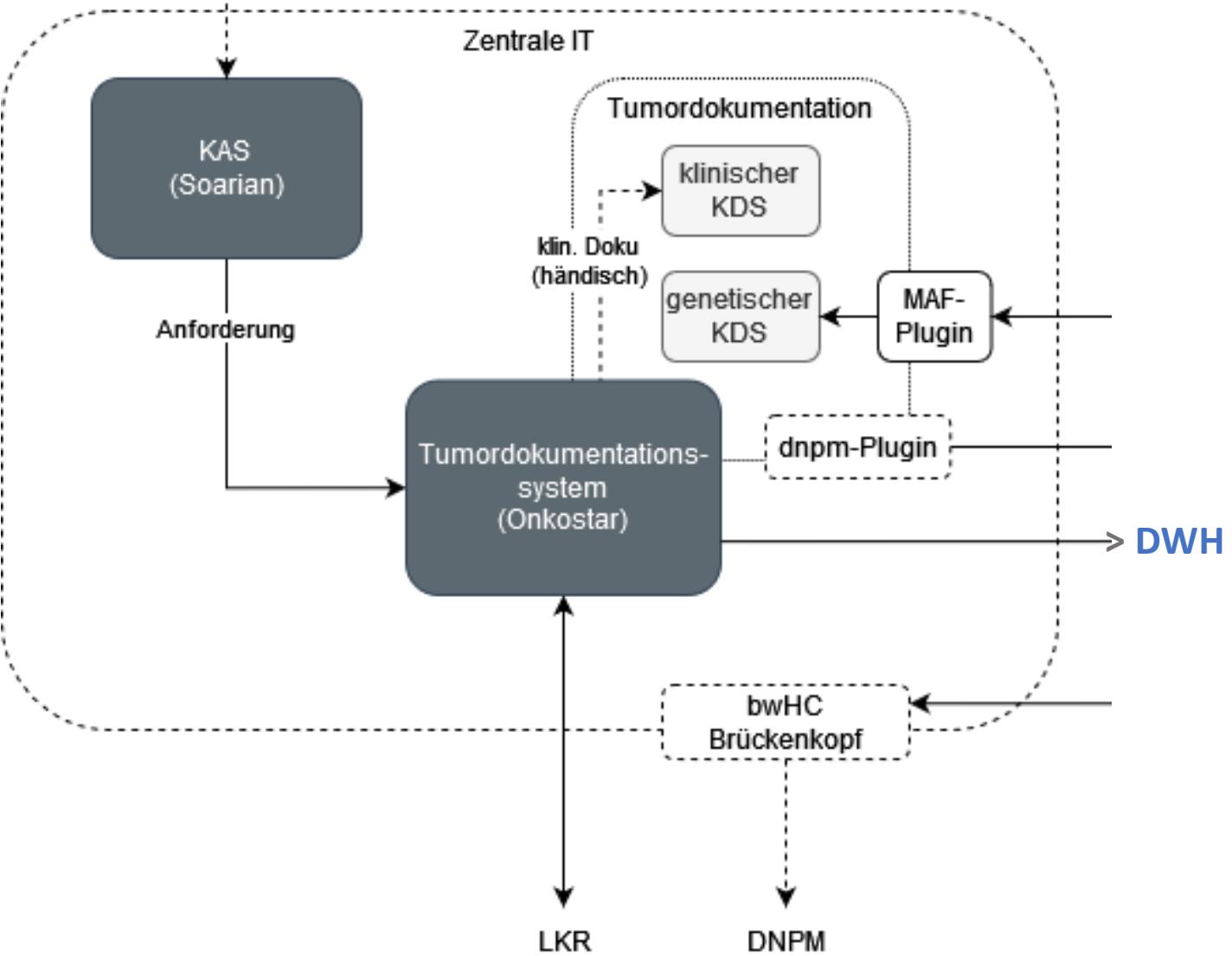
benötigt .maf-Datei

- aus vcf generiert
- mehr Informationen als Excel

→ Ausleitung ins Data Warehouse



# Einbindung in die IT-Landschaft - Detailansicht Onkostar



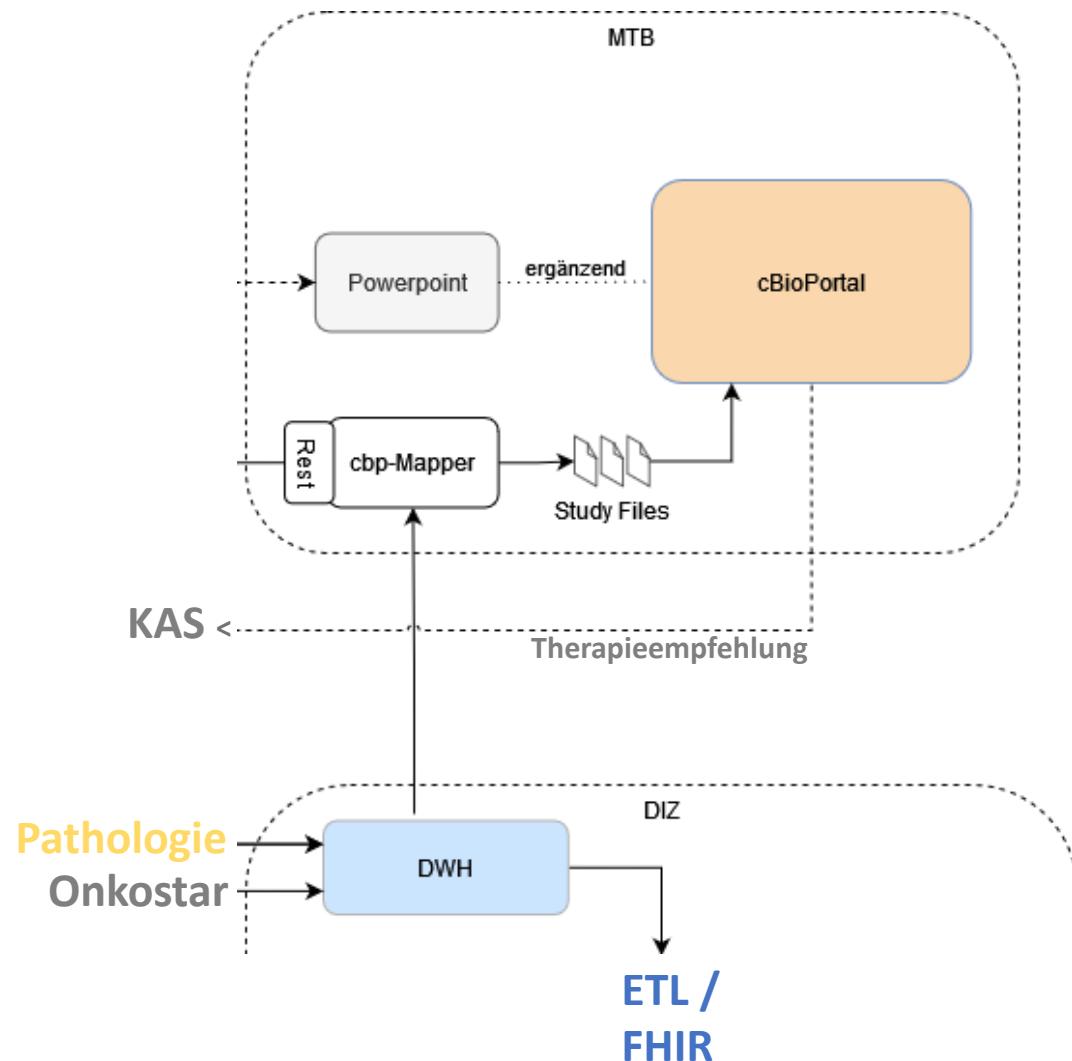
## Onkostar

- Dokumentation der klinischen Daten (Diagnosen, Verlaufsdaten, etc...) via DNPM-Formulare
- relevante klinische Informationen fürs MTB werden über DB-View bereitgestellt

→ Ausleitung ins Data Warehouse

- genetischer KDS wird über REST-API mit Daten aus Pathologie ergänzt

# Einbindung in die IT-Landschaft - Detailansicht cBioPortal



## Molekulares Tumorboard

- nutzt immer noch primär PowerPoint
- cBioPortal ergänzend (auch in Vor-/Nachbereitung der Fälle)

## Befüllung cBioPortal

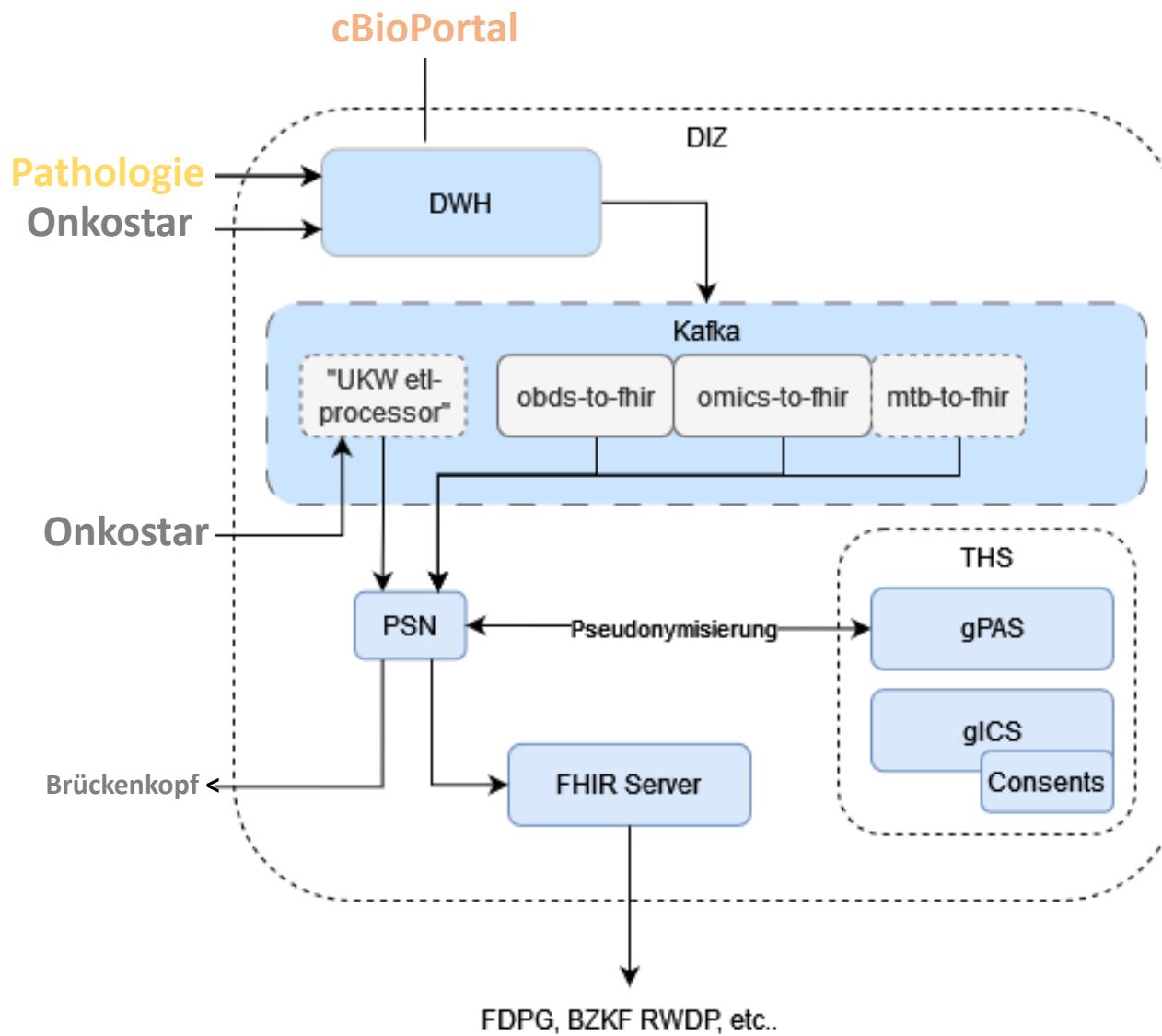
- über „cbpMapper“ (Eigenentwicklung)
- erzeugt Study Files (txt-Dateien) für den Import nach cBioPortal

## Therapieempfehlung

- bisher noch nicht direkt in cBioPortal erfasst



# Einbindung in die IT-Landschaft - Detailansicht DIZ



## Ausleitung in FHIR

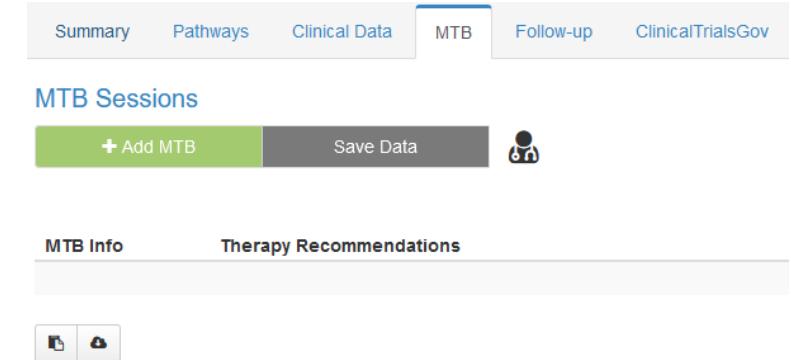
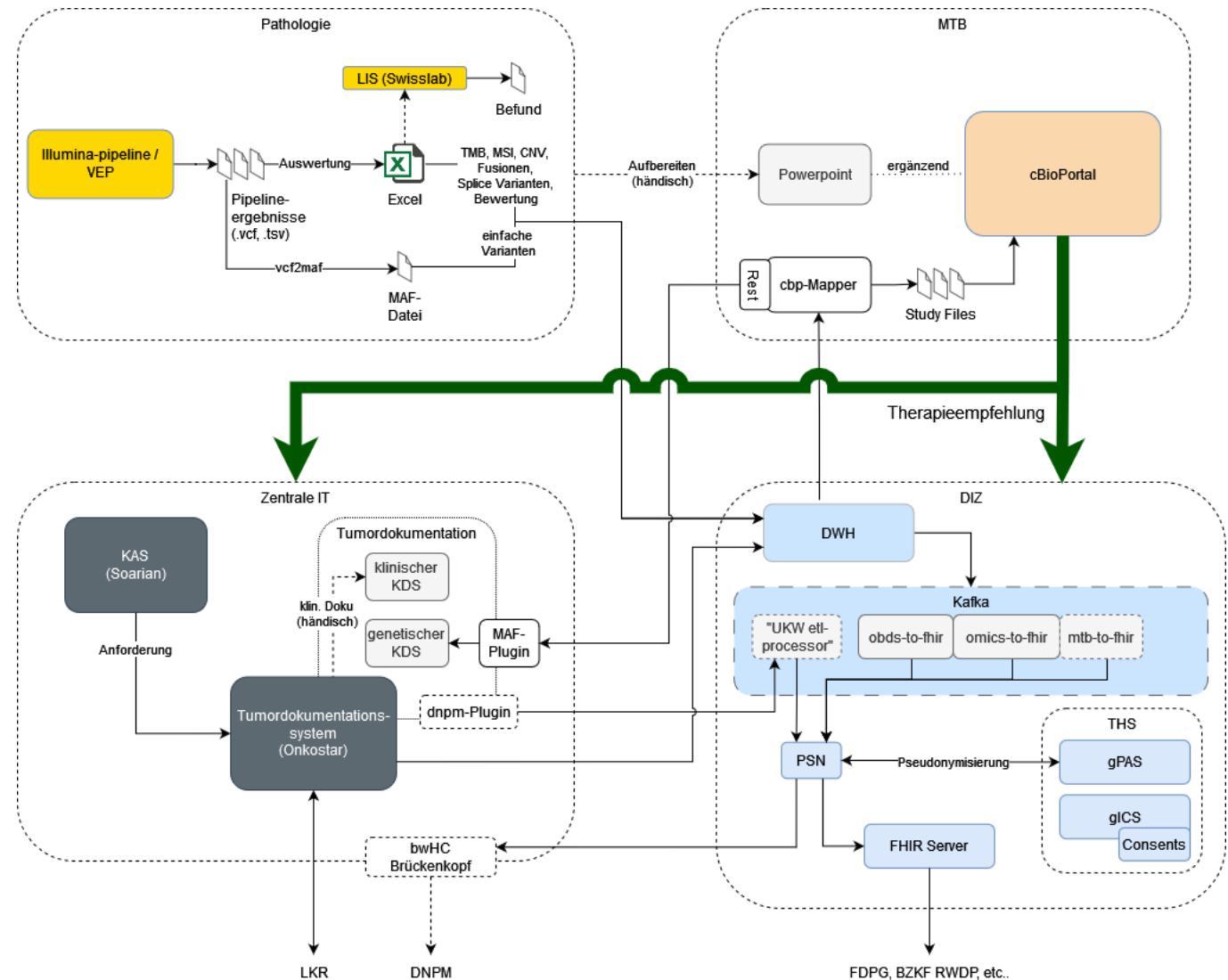
- Modul Onkologie
- Modul MolGen Befundbericht

zukünftig auch:

- Modul molekulares Tumorboard
- (Pathologiebefund)
- Ausleitung über Brückenköpfe für andere Projekte wie z.B. DNPM

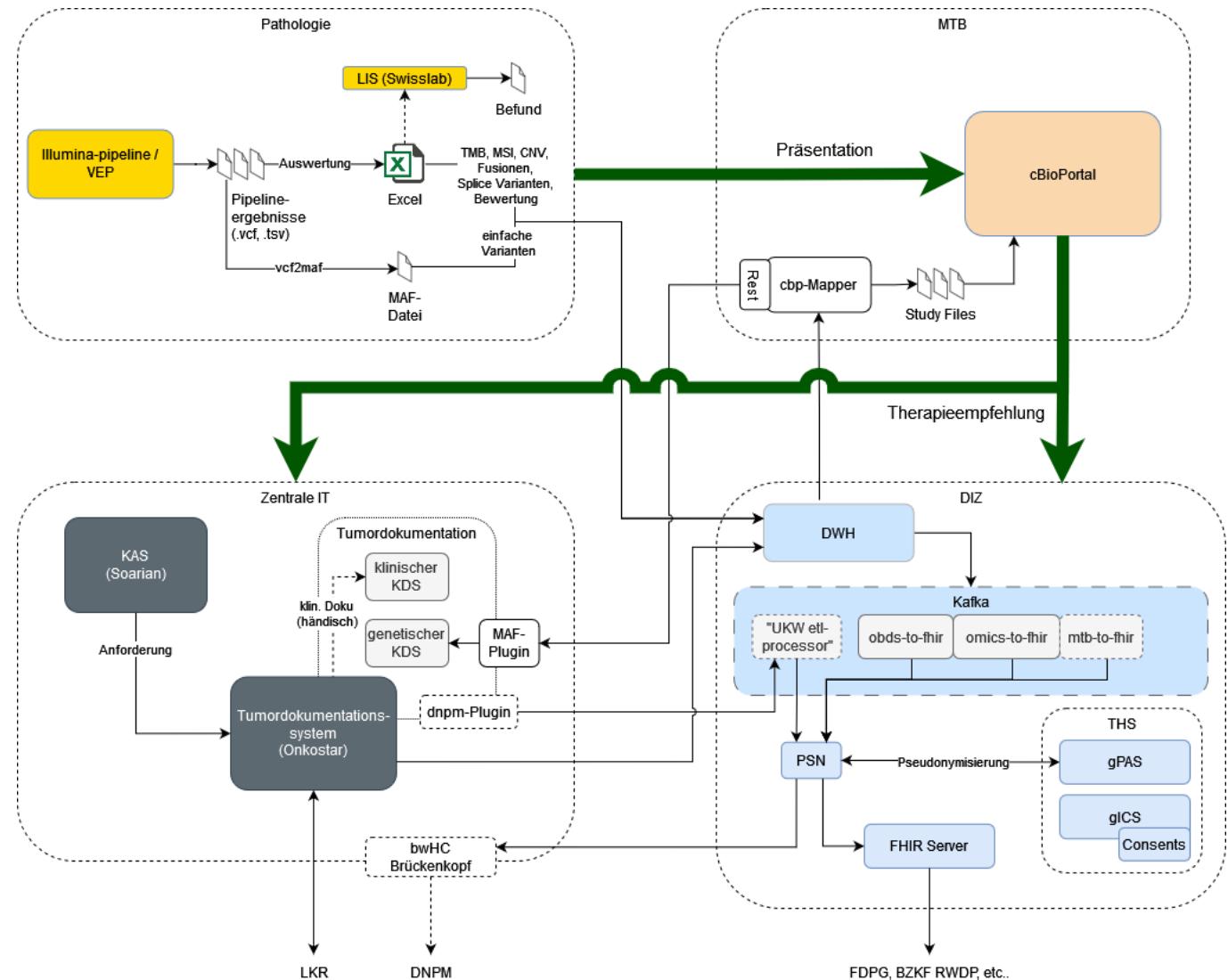


# Einbindung in die IT-Landschaft - Erweiterungen





# Einbindung in die IT-Landschaft - Erweiterungen



The mockup shows the MTB presentation interface with the following sections:

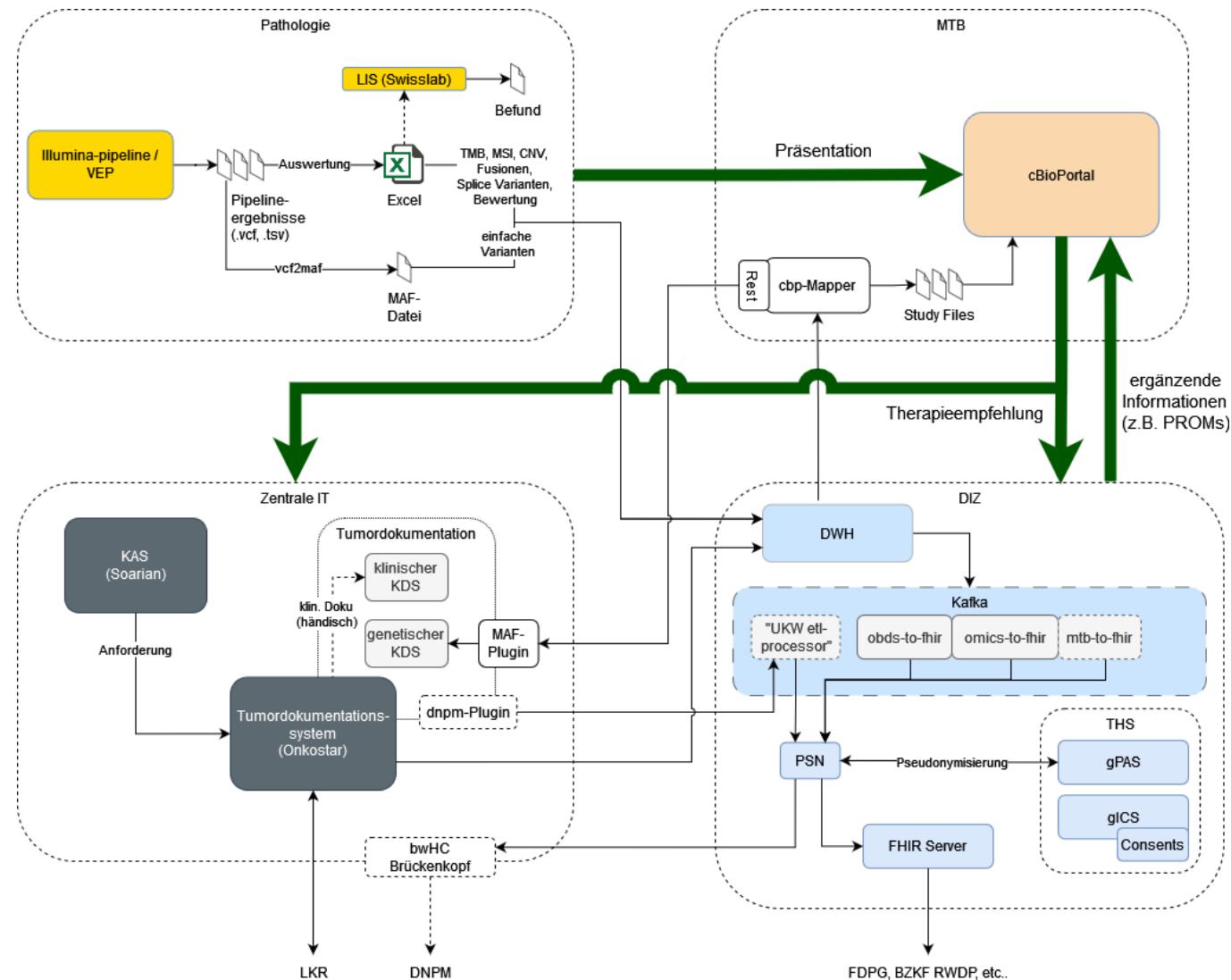
- MTB Sessions:** Includes buttons for "+ Add MTB" and "Save Data".
- MTB Info:** Placeholder for MTB information.
- Therapy Recommendations:** Placeholder for therapy recommendations.
- Summary, Pathways, Clinical Data, MTB, Follow-up, ClinicalTrialsGov:** Navigation tabs.
- MTB Presentation:** Placeholder for the presentation slide.
- Add:** A modal dialog with options: Molecular Findings, Medical history, Clinical Data, and Diagrams.
- Beschreibung der nicht-sy Gen:** A table showing genetic variants:
 

Gen	Exon	HSI
KMT2C	34/59	c.5
BRCA1	15/18	c.1
JAK1	26/27	c.3
FOXA1	2/2	c.11

Quelle: Marco Schindler – Mockup Einführungsvortrag 21.05.2024



# Einbindung in die IT-Landschaft - Erweiterungen



The mockup shows the MTB presentation interface with the following sections:

- Header:** Summary, Pathways, Clinical Data, MTB (selected), Follow-up, ClinicalTrialsGov
- MTB Sessions:** Add MTB, Save Data
- MTB Info:** Therapy Recommendations
- Content:** A detailed view of a therapy recommendation for a patient named "Bachmeier, Horst geb. 03.02.1938". The recommendation includes information about the tumor (Lympho-Plasmazelluläre Kryptome, ED 03/2019), treatment (Bachmeier, Horst geb. 03.02.1938), and side effects (e.g., Diarrhoe, 2x/Tag). A sidebar shows an "Add" button with options: Molecular Findings, Medical history, Clinical Data, and Diagrams.
- Bottom:** Beschreibung der nicht-sy Gen, KM12C, BRAF, JAK1, FOXA1; Exon, 34/59, 15/18, 26/27, 2/2; c.5, c.1, c.3, c.11.

Quelle: Marco Schindler – Mockup Einführungsvortrag 21.05.2024



# Vielen Dank für Ihre Aufmerksamkeit!



@miracum\_de



@DIFUTUREde



GEFÖRDERT VOM



Bundesministerium  
für Bildung  
und Forschung