

Transforming Precision Diagnostics in Switzerland

Applying Whole Genome and Whole Transcriptome Sequencing for Improved Patient Outcomes in Cancer Care

Clinical solid tumor oncology

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Whole genome sequencing (WGS) has been validated technically and clinically by comparing its performance to established pan-cancer (Foundation One CDx, 324 genes) and melanoma-specific (190 genes) panels⁴. WGS demonstrated analytical validity, by detecting up to 98% of classic mutations and markers. In addition, WGS also identified complex biomarkers such as UV-associated mutational signatures, HLA types, and genome-wide copy number alterations, broadening its clinical utility. The completeness of WGS data further enables its use in reporting pharmacogenomic mutations⁵, chimerism, and tissue typing, offering long-term utility as compared to gene panels.

Additionally, metagenomic WGS pipelines have been established for analysis of FFPE samples⁶. In 2022, Swiss pathologists developed a metagenomics-based pipeline that successfully identified an infectious pathogen as the underlying cause of a suspected lung tumor.

WGS is improving patient care by delivering an interdisciplinary wholistic molecular view on the patient with a single analysis. If properly set up, this one-stop-shop type of analysis will save time and may dramatically alter the frequency of unexpected, but actionable feedback.

⁴Litchfield, C. & Nienhold, R et al, Integrating FFPE derived Whole Genome Sequencing into Routine Molecular Pathology. unpublished
⁵Swen J.J. et al, A 12-gene pharmacogenetic panel to prevent adverse drug reactions. The Lancet 2023
⁶Nienhold, R. et al, Unbiased screen for pathogens in human FFPE samples by WGS and metagenomics. Front Cell Infect Microbiol 2022

Complex biomarkers
(Molecular pathology
& molecular hematology)

MSI

Pattern & signature
recognition

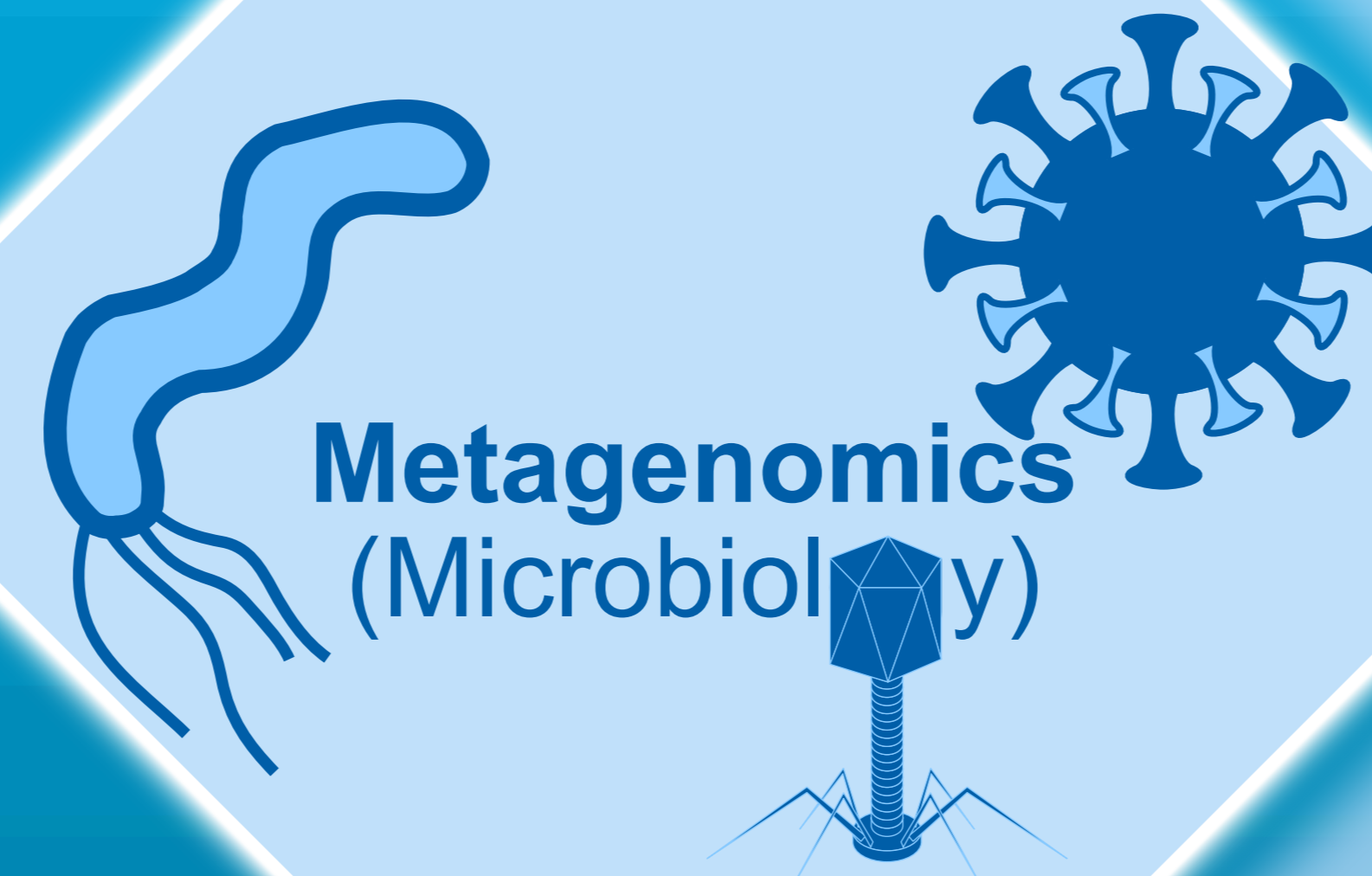


Somatic alterations
(Molecular pathology
& molecular hematology)

SNV

FUSION

CNV

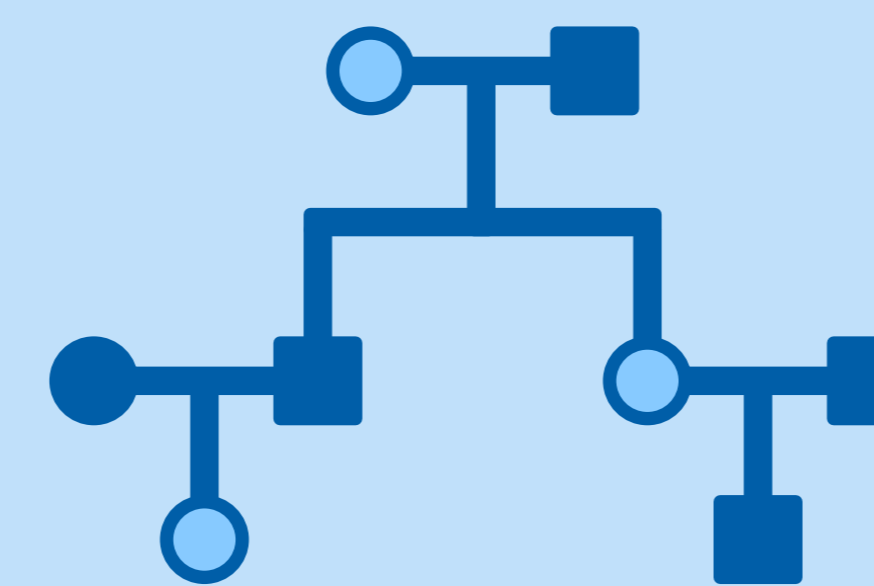


Metagenomics
(Microbiology)

WGS is improving patient care by delivering an interdisciplinary wholistic molecular view on the patient with a single analysis. If properly set up, this one-stop-shop type of analysis will save time and may dramatically alter the frequency of unexpected, but actionable feedback.

WGS

Germline alterations
(Medical genetics)



Blood & HLA typing
(Immunology)



HLA-A, -B, ...

A, B, 0
+/-

Pharmacogenomics
(Toxicology)

	safety-code The Medication Safety Code (MSC) is a patient-specific code on the left represents a patient-specific genetic profile regarding important pharmacogenes.	safety-code Name: Jane Doe Date of birth: 01.02.1994 Critical drug-substance (modification recommendation): CYP2D6 Clozapine, Sertraline
	How does it work? After scanning the QR code (e.g. with a smartphone), you are led to a website that displays patient-specific drug dosing recommendations.	Other genes: ANKK1, ADORA1, BRCA1, CYP1A1, CYP2A6, CYP2B6, CYP2C9, CYP2A4, CYP2A5, CYP2A6, CYP2A7, CYP2A8, CYP2A9, CYP2A10, CYP2A11, CYP2A12, CYP2A13, CYP2A14, CYP2A15, CYP2A16, CYP2A17, CYP2A18, CYP2A19, CYP2A20, CYP2A21, CYP2A22, CYP2A23, CYP2A24, CYP2A25, CYP2A26, CYP2A27, CYP2A28, CYP2A29, CYP2A30, CYP2A31, CYP2A32, CYP2A33, CYP2A34, CYP2A35, CYP2A36, CYP2A37, CYP2A38, CYP2A39, CYP2A40, CYP2A41, CYP2A42, CYP2A43, CYP2A44, CYP2A45, CYP2A46, CYP2A47, CYP2A48, CYP2A49, CYP2A50, CYP2A51, CYP2A52, CYP2A53, CYP2A54, CYP2A55, CYP2A56, CYP2A57, CYP2A58, CYP2A59, CYP2A60, CYP2A61, CYP2A62, CYP2A63, CYP2A64, CYP2A65, CYP2A66, CYP2A67, CYP2A68, CYP2A69, CYP2A70, CYP2A71, CYP2A72, CYP2A73, CYP2A74, CYP2A75, CYP2A76, CYP2A77, CYP2A78, CYP2A79, CYP2A80, CYP2A81, CYP2A82, CYP2A83, CYP2A84, CYP2A85, CYP2A86, CYP2A87, CYP2A88, CYP2A89, CYP2A90, CYP2A91, CYP2A92, CYP2A93, CYP2A94, CYP2A95, CYP2A96, CYP2A97, CYP2A98, CYP2A99, CYP2A100

CYP2D6*4