

# **Roche Solutions for Sequencing**

Our vision: A future where NGS is simple and accessible enough for routine clinical use



# **Research Focus**

Oncology Molecular profiling ctDNA analysis Hereditary diseases Genomic biomarkers Cardiology Neurology Infectious diseases

# **Workflows & Methods**

Whole genome sequencing (WGS) Whole exome sequencing (WES) RNA sequencing (RNA-seq) Targeted sequencing (DNA, RNA) NGS library prep automation Digital PCR (dPCR) and real-time PCR/qPCR FFPET sample dissection Automated nucleic acid extraction



# Solutions for Sequencing



# More Sequencing Solutions

#### **Custom Sample Prep Solutions**

Collaborative creation of unique workflows and kits with experts at the Roche Support Network.

Page 13

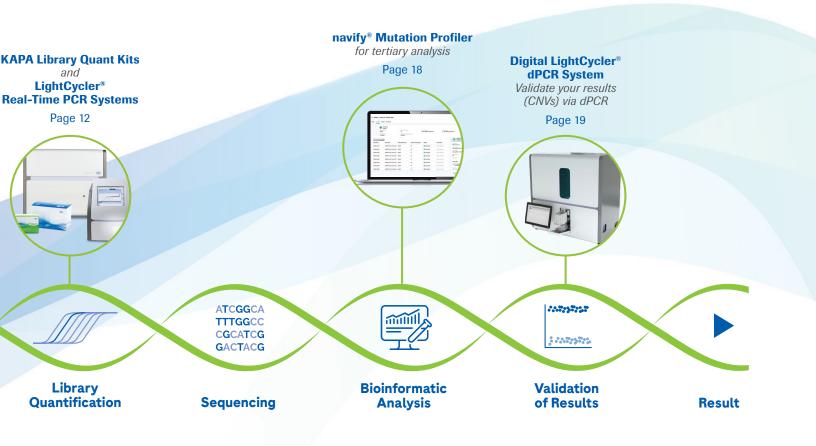


#### AVENIO Edge Workstation or automation on non-Roche systems

Fully automated system for library preparation, target enrichment, pooling, normalization, and quantification.

Pages 14, 15





#### **AVENIO ctDNA & Tumor Tissue Analysis Kits**



#### **AVENIO Tumor Tissue CGP Kit**



The MagNA Pure 24 and MagNA Pure 96 Systems, AVENIO Edge Workstation, and the Digital LightCycler® dPCR System are IVD products. The workflow above shows these products being utilized within a research workflow with other Roche products that are Research Use Only.

Because every sample is precious, continued support across the NGS workflow is critical.

# Support for the development and validation of seamless workflows

to achieve the sensitivity and precision expected from the world's largest in vitro diagnostics company





# **Products designed to work together across the entire NGS workflow**—built

upon decades of technology leadership in NGS, target enrichment, and sample prep automation

# A single source for technical expertise for all aspects of library preparation,

from ideation through ongoing support—ensuring reliable, reproducible, high-quality results for assay after assay after assay





# Nucleic Acid Extraction MagNA Pure 24 and MagNA Pure 96 Systems

**High-quality starting material increases sequencing success.** High-molecularweight input DNA is essential for the creation of libraries with the 350 – 650 bp inserts required for sequencing whole human genomes on Illumina<sup>®</sup> NovaSeq,<sup>™</sup> NextSeq, and HiSeq<sup>®</sup> instruments.

Obtain high-quality, high-molecular-weight DNA for direct use in sequencing with the **MagNA Pure 24** and **MagNA Pure 96 Systems**. These fully automated nucleic acid extraction instruments provide walkaway automation, require less user intervention, and minimize variability between extractions.

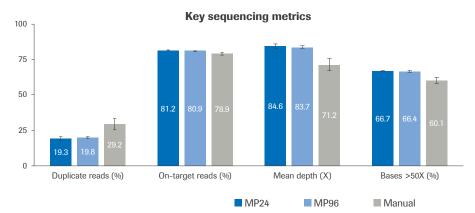
- Reliable DNA extraction from as little as 200 µL whole blood
- Scalable extraction for low, mid, or high throughput
- Optimized protocols for NGS workflows with blood, plasma, or FFPET samples
- Superior sequencing coverage and fewer duplicate reads compared to manual extraction (**Figure 3**)

#### MagNA Pure 24 System









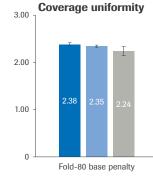


Figure 3. cfDNA extracted from MagNA Pure 24 and MagNA Pure 96 Systems yields superior sequencing coverage and fewer duplicate reads compared to manual extraction. Target-enriched libraries were prepared from cfDNA isolated from 4 mL of plasma, using either the MagNA Pure 24 cf ds 4000 hp protocol, the MagNA Pure 96 cfdna ds 4000 protocol, or a manual protocol. The total yield of cfDNA for each sample was used as input into the HyperCap 2.0 workflow, using the KAPA HyperPrep Kit and the SeqCap EZ Human Oncology Panel (2.75 Mb). Prior to target capture, libraries were multiplexed; each capture contained samples obtained from each of three extraction workflows. Sequencing was performed on the NextSeq 500 (2 x 75 bp). Raw reads were randomly downsampled to 6M prior to analysis. Each bar represents the mean of 5 replicate extractions; error bars indicate the standard deviation.

#### Table 1. Overview of NGS-compatible MagNA Pure System workflows.

Platform	Sample input	Nucleic acids output	Protocol
	blood	genomic DNA	hgDNA ds 200
MagNA Pure 24	plasma	cell-free DNA	cfNA ds 4000 hp
	FFPET	FFPET DNA	DNA FFPET 1000
MagNA Pure 96	blood	genomic DNA	DNA Blood ds SV





# Sample Quantification / QC Roche LightCycler<sup>®</sup> 480 System and KAPA NGS FFPE QC Kit

The amount of intact DNA that is used as input into DNA library preparation can have a big impact on the molecular complexity of the resulting libraries; this, in turn, can determine whether the libraries will yield the desired sequencing coverage.

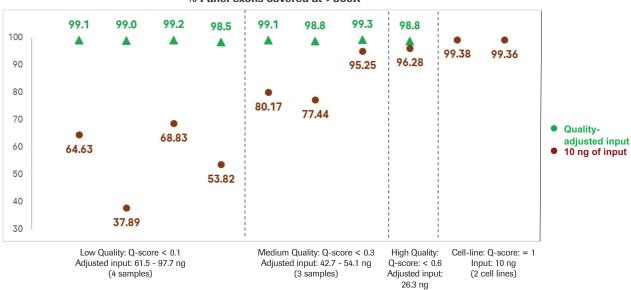
qPCR-based QC of input DNA provides the most accurate assessment of the quality of the input DNA, providing the user with data that enables them to optimize input amounts based on sample quality.

KAPA NGS FFPE QC Kits determine sample guality using two sets of primers that target the human Long Interspersed Nuclear Elements (LINE), which occur across the genome. Amplification with these primers yields a shorter amplicon (66 bp) and longer amplicon (191 bp). The ratio of these two products is used to calculate the Q-score and adjust input amounts, enabling the user to:

- Improve library quality and sequencing coverage from low- or medium-quality samples.
- Assess input guality with greater accuracy compared to assays of individual housekeeping genes.
- · Conserve sample when using high-quality samples.



The Roche LightCycler<sup>®</sup> 480 System ensures reproducible, reliable, accurate data. Leverage the dependable accuracy and temperature homogeneity of the LightCycler® 480 System with KAPA NGS FFPE QC Kits.



% Panel exons covered at >300X

Figure 4. Low-quality, medium-quality, and high-quality input DNA yielded comparable results when quality-adjusted input amounts were used in the KAPA HyperPETE Somatic Tissue DNA Workflow. Designed and supported for the KAPA HyperPETE Somatic Tissue DNA Workflow. Contact US Support & Applications for guidance with other applications and instruments.



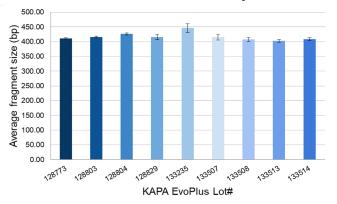
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**KAPA DNA library prep kits** deliver high performance across a diverse array of experimental conditions and sample inputs. Workflows are made simple with sample-conserving, automatable protocols that free up valuable hands-on time while delivering reproducible, high-quality results. Choose from PCR-free or with-PCR formats that include our low-bias, high-fidelity KAPA HiFi DNA polymerase for mechanical or enzymatic fragmentation methods.

- Rely on consistent performance of kits manufactured to meet the highest quality standards.
- Save precious sample and hands-on time with streamlined workflows.
- Preserve library diversity with minimal bias for a seamless transition into downstream applications.

Minimal lot-to-lot variabilty



**Figure 5. KAPA EvoPlus Kits' performance is highly consistent across lots due to Roche's high quality standards.** This data was generated from 8 lots of KAPA EvoPlus Kits using 100 ng input *E. coli* DNA fragmented for 15 mins. Adapter ligation was performed using 10µM KAPA UDI Adapter with a 0.8X cleanup using HyperPure beads post ligation. Fragment size was assessed on an Agilent Bioanalyzer (High Sensitivity assay).

# Which kit is right for you?

#### **KAPA HyperPrep Kits**

Streamlined, sample-sparing workflow delivers superior low-bias results for mechanically sheared inputs, including cfDNA.

### **KAPA HyperPlus Kits**

Single-tube, enzymatic fragmentation is a robust solution for applications requiring the highest yields from low-quality sample inputs such as FFPE.

### **KAPA EvoPlus Kits**

Novel, ReadyMix workflow with enzymatic fragmentation consistently delivers unparalleled convenience, remarkable inhibitor tolerance, and high performance with WGS sample inputs.



#### Table 2. Comparison of KAPA DNA library preparation kits

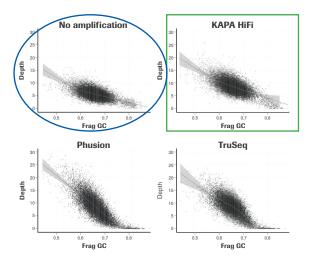
	KAPA HyperPrep Kits	KAPA HyperPlus Kits	KAPA EvoPlus Kits
Fragmentation method	Mechanical	Enzymatic	Enzymatic
Hands-on time	Good	Better	Best
Inhibitor tolerance	Robust	Low; EDTA-sensitive	Broad tolerance, including EDTA
Sample input range	1 ng - 1 µg	1 ng - 1 µg	10 ng - 500 ng
PCR and PCR-free formats	8, 24, 96 rxn tubes	8, 24, 96 rxn tubes	24, 96, 384 rxn tubes & 96 rxn plates

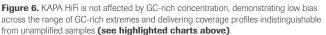


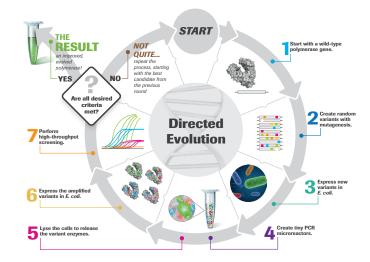


**KAPA Library Amplification Kits** harness the performance benefits of KAPA HiFi DNA Polymerase—a novel, broadly referenced enzyme specially formulated to minimize amplification bias while maintaining extremely high fidelity. KAPA Library Amplification Kits can be used with usersupplied primers or with our KAPA Library Amplification Primer Mix, which contains primers complementary to Illumina sequencing adapters.

- Improve the detection of true and rare variants with high fidelity.
- Achieve uniform coverage of low-GC/AT-rich regions with low bias.
- Accurately amplify long, complex, and high-GC-content templates with superior processivity.







Typically, choosing a DNA polymerase requires trade-offs between essential performance characteristics such as fidelity or the amplification of long and difficult targets, or even settling for compromised performance at application-specific experimental extremes such as inhibitor concentration. Referenced in thousands of scientific publications, KAPA HiFi DNA Polymerase is a product of Directed Evolution—an iterative and highly selective enzyme synthesis process that enables the blending of critical performance parameters and the precise selection of enzymes that deliver uncompromising performance over the full spectrum of experimental conditions.

# **KAPA Library Amplification Kits and Primer Mixes**

# KAPA Library Amplification & HiFi ReadyMix Kits

For low-bias production of sequencing-ready libraries

# KAPA Library Amplification Kits with Primer Mix

With primers complementary to the P5 and P7 regions of Illumina adapters and indexing primers

### KAPA Library Amplification Primer Mix

Specially formulated to limit primer depletion and over-amplification, and available in a convenient 96-rxn plated format



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RNA library preparation is the critical first step of RNA sequencing (RNA-seq). **KAPA RNA HyperPrep Kits** offer a wide variety of streamlined workflow options for single-tube, single-day library prep that include enrichment of target transcripts by selective mRNA capture or rRNA depletion.

All KAPA RNA HyperPrep Kits contain high-quality buffers and enzymes—including KAPA HiFi DNA Polymerase, developed through our Directed Evolution Technology—for the preparation of RNA libraries that yield minimal GC bias and uniform sequence coverage.

- Create high-quality libraries that yield high-quality data, even with degraded and low-input samples.
- · Deplete custom-selected RNA targets, in addition to rRNA and globin transcripts.
- Automate KAPA RNA HyperPrep on a wide variety of liquid handlers (see page 15).

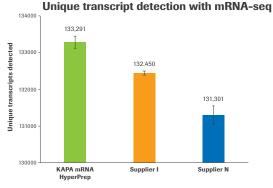


Figure 7. Efficient mRNA capture using KAPA mRNA HyperPrep leads to the detection of more unique transcripts vs. other suppliers. Libraries were generated in quadruplicate using Universal Human Reference (UHR) RNA (Agilent Technologies).

#### Table 3. Overview of KAPA RNA library preparation kits

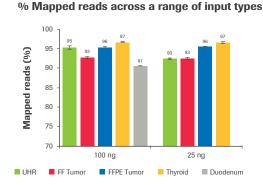


Figure 8. KAPA RNA HyperPrep yields over 90% mapped reads, even with low inputs of FFPET RNA. RNA samples of varying quality were used as input into library construction using KAPA RNA HyperPrep Kit with RiboErase (HMR) with 25 and 100 ng inputs. For highly degraded duodenum FFPE RNA, libraries were only prepared with 100 ng input. All samples were prepared using the standard adapter stock concentration (1.5  $\mu$ M) and post-ligation cleanup ratios (0.63X/0.7X).

	KAPA RNA HyperPrep Kits	KAPA RNA HyperPrep Kits with RiboErase (HMR)	KAPA RNA HyperPrep Kits with RiboErase (HMR) Globin	KAPA mRNA HyperPrep Kits
<b>RNA enrichment</b>	None	rRNA depletion	rRNA and globin depletion	Poly(A) selection
Sample type	<ul> <li>High-quality total RNA</li> <li>Degraded or FFPE total RNA</li> <li>Previously enriched RNA</li> </ul>	<ul> <li>High-quality total RNA</li> <li>Degraded or FFPE total RNA</li> </ul>	<ul> <li>Blood-derived RNA</li> <li>High-quality total RNA</li> <li>Degraded or FFPE total RNA</li> </ul>	High-quality total RNA
Species	<ul> <li>Eukaryotic (animal, plant, etc.)</li> <li>Prokaryotic (bacterial, etc.)</li> </ul>	- Human, mouse, and rat*	<ul> <li>Human, mouse, and rat*</li> </ul>	<ul> <li>Eukaryotic (animal, plant, etc.)</li> </ul>
Differentiating applications	Analysis of specific transcripts, including those of low abundance, when paired with target enrichment	Whole transcriptome analysis, including non-coding RNA profiling	Whole transcriptome analysis, including non-coding RNA profiling	mRNA sequencing for coding transcriptome analysis
Shared applications	Gene expression analysis; detection	n of gene fusions, isoforms, and other	r structural variants; SNV discovery	
Shared highlights	Streamlined and automation friendlor mRNA enrichment	y; low duplication rates and high cove	erage uniformity; single-day workflow	s including depletion

\*Custom depletion protocol support available for other organisms or transcripts.





**Target Enrichment** KAPA HyperCap Probes for Hybridization-based Target Enrichment

## **Better by Design**

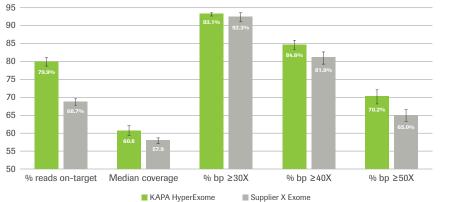
Combining nearly two decades of probe-design experience with an improved manufacturing process, KAPA HyperCap Probes offer fully customizable target enrichment panels and pre-designed panels for hybridization-based capture before next-generation sequencing, KAPA HyperCap Probes are manufactured using KAPA HiFi DNA Polymerase and are validated by NGS, resulting in high-quality probes designed to answer your most challenging research questions.

Ready-to-ship, predesigned KAPA HyperCap Fixed Panels enable faster access to relevant content, and include KAPA HyperExome, KAPA HyperCap Oncology Panel, and KAPA HyperCap Heredity Panel. Additional designs are available from our KAPA HyperCap Design Share Panel collection-developed in collaboration with leading researchers around the world—and include panels for hereditary conditions, oncology, and metabolic disease research.

Custom target enrichment panels are easily designed using HyperDesign online software (see below). HyperDesign can be used to create either human designs (KAPA HyperChoice Probes) or nonhuman designs (KAPA HyperExplore Probes).

### Combine KAPA HyperCap Probes with KAPA Library Preparation Kits to:

- Reduce sequencing costs and save time with superior capture uniformity.
- Reliably enrich challenging, previously inaccessible genomic regions.
- Streamline targeted enrichment using the updated, readily automated KAPA HyperCap Workflow, driven by KAPA HyperPrep or KAPA HyperPlus Library Prep Kits.



#### Key enrichment and sequencing metrics

#### Figure 10. KAPA HyperExome yields greater % reads-on-target, deeper median coverage, and broader target coverage compared to the Supplier X exome. DNA from 16 cell lines was processed in triplicate (48 libraries per workflow); input DNA was enzymatically sheared; samples were pre-capture multiplexed in sets of 8 and hybridized for 16 hours; final post-capture libraries were amplified with 8 PCR cycles: and libraries were sequenced (2 x 100 bp) on an Illumina® NovaSeq<sup>™</sup> sequencer. For analysis, sequencing data was subsampled proportionally to exome panel size to achieve the same targeted average depth of coverage.

# Reliably enrich challenging, previously inaccessible genomic regions

The user-friendly, online HyperDesign tool builds on two decades of in silico design experience to select probe panels that achieve deeper, more uniform downstream sequencing coverage with fewer sequencing reads-even across difficult-to-capture regions.

Design your new probe panel in 4 easy steps:

- 1. Visit www.HyperDesign.com and select your organism of interest;
- 2. Add your targets by uploading gene names, bed files, or genomic coordinates-or choose from a broad list of commonly used gene identifiers;
- 3. Fine-tune your inputs, review your targets, and confirm your results; and
- 4. Submit your design for probe selection.





HyperDesig

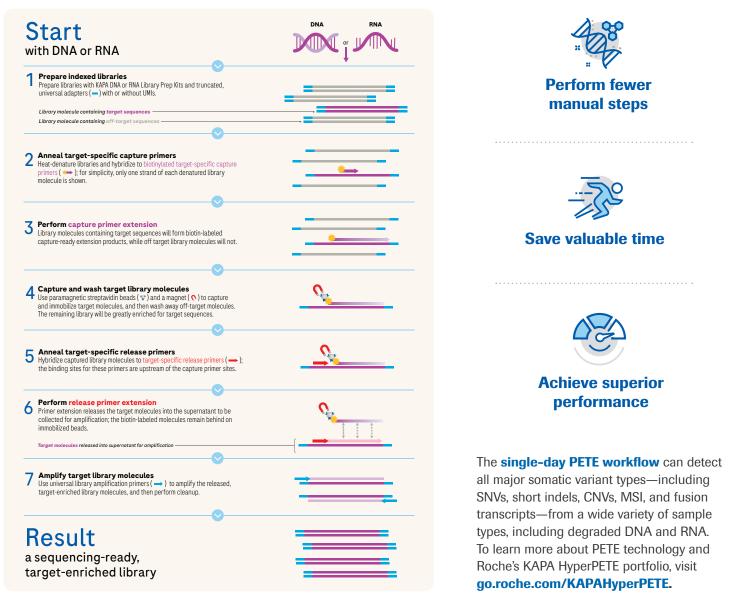


PETE is a novel NGS hybridization capture technology designed to employ primer extension reactions to specifically capture and release target library molecules for sequencing.

# What's different about PETE?

Other target enrichment technologies offer either uniform, high-quality data (via probe hybridization) or fast, simple workflows (via amplicon-based enrichment). PETE brings together the benefits of both workflows—combining speed and simplicity with deep, uniform, high-quality coverage.

# Here's how PETE works...







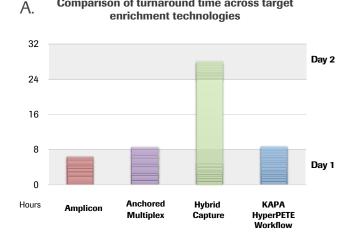
# Target Enrichment KAPA HyperPETE Primers for Primer Extension Target Enrichment

## Combine the performance of hybrid-capture target enrichment with the speed and simplicity of amplicon workflows

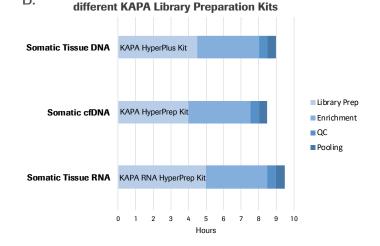
KAPA HyperPETE is a novel hybrid-capture technology designed to employ primer extension reactions to specifically capture and release target library molecules for sequencing. It is designed and optimized to detect all major somatic variant types, including SNVs, short indels, CNVs, MSI, and fusion transcripts (known and novel). KAPA HyperPETE is compatible with a wide variety of sample types, including challenging samples-such as cfDNA and FFPET-derived DNA and RNA.

The KAPA HyperPETE Portfolio includes readily available fixed-design panels for hereditary oncology, oncology hotspots, lung cancer fusion variants, and pan-cancer variants (with an MSI module). In addition, custom panels can be designed using HyperDesign, our easy-to-use online design tool.

- Save valuable time with an efficient, single-day (less than 9 hours), automation-friendly workflow.
- Achieve superior performance and coverage uniformity.
- Uncover critical genomic information from a wide variety of sample types, including FFPET and cfDNA.
- Take hours off of total workflow time compared to typical hybridization capture, with time requirements similar to amplicon and anchored multiplex methods (Figure 9)
- Enrich for long contiguous regions, using fewer tubes per sample compared to amplicon workflows.



Comparison of turnaround time across target



KAPA HyperPETE turnaround times using three



Figure 9. The turnaround time (TAT) for KAPA HyperPETE target enrichment is similar to the TAT for amplicon-based workflows. (A) While most hybridization-based workflows take two days to complete, KAPA HyperPETE workflows can be completed in one day. (B) Differences in the TAT for various applications of KAPA HyperPETE are dependent on the library preparation kit used, as each kit requires slightly different completion times. However, once the libraries are created, the enrichment workflow is the same across applications.

B.





Sequencing capacity is maximized when sequencing-competent molecules are accurately measured with qPCR, enabling libraries to be pooled at the desired ratios.

Clustering can be optimized by quantification of library pools, further improving sequencing results.

Roche LightCycler<sup>®</sup> 96 Instrument and LightCycler<sup>®</sup> 480 System ensure reproducible, reliable, accurate data.

- Scalable instrument options
- Dependable temperature accuracy and homogeneity
- Ideal for use with KAPA Library Quantification Kits

**KAPA Library Quantification Kits**, which are referenced in thousands of scientific publications, contain all reagents needed for qPCR-based quantification of NGS libraries for Illumina<sup>®</sup> sequencing.

- Accurately quantify sequencing-competent libraries (Figure 11).
- Pool libraries with better accuracy for more balanced multiplexing.
- Automate KAPA Library Quantification Kits for increased throughput (see page 15).





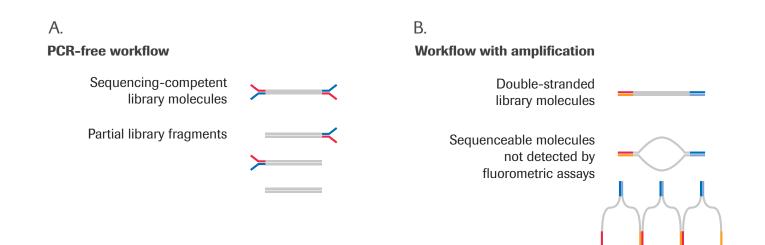


Figure 11. Library quantification via qPCR-based methods, such as the KAPA Library Quantification Kit, enables accurate sample pooling and optimal clustering. (A) Libraries prepared with PCR-free workflows can contain partial library fragments that are not sequenceable. qPCR-based library quantification methods detect

only the sequencing-competent molecules. In contrast, other assays detect fragments that are not sequenceable, leading to *underclustering* on the sequencing flow cell.

(B) Libraries prepared using methods with PCR amplification can include sequencing-competent single-stranded configurations. qPCR-based library quantification data counts these molecules. In contrast, other methods do not detect these molecules, leading to *overclustering* on the sequencing flow cell.



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# **Custom Sample Prep Solutions**

Unique kits designed for your unique workflows

Created with the same high-quality **KAPA library prep reagents** in combinations that help you maximize efficiency, increase throughput, and reduce costs.





Preparation



Target Enrichment



Sample Quantification/QC

Automation

Collaborative creation of robust workflows with experts at the Roche Support Network







NGS Sample Prep Automation

Maximize the potential of your lab with a trusted NGS partner

Automate your medium-throughput NGS library preps with as little as 20 minutes of hands-on time and no automation experience.

Freedom to walk away with confidence and trust in the results **AVENIO Edge System** is Roche's fully automated solution for NGS library preparation, including target enrichment and library pooling. It is designed to greatly reduce the complexity of automation for users at any level, and provide a true walk-away experience.



For more information about the AVENIO Edge System, contact your Roche sales representative or visit:

sequencing.roche.com/AVENIOEdge





# Automated KAPA NGS Workflows

For non-Roche liquid handlers



# Thinking of automating library prep on your existing liquid handler?

**Roche's NGS Automation Support Team** can help develop automated KAPA Library Prep protocols as a complimentary service for our customers, assisting you at every step of the way.

Let's talk about the next steps to get you up and running with your methods.

Scan the QR barcode below or fill out the Contact Us form at **sequencing.roche.com/NGSAutomation**.

	ndor atform	DNA Library Preparation			RNA Library Preparation				Quant and QC	Target Enrichment	Oncology	
Vendor	Platform	Kapa	KAPA	KAPA	EvoPlus	KAPA Stranded	KAPA Total RNA	KAPA mRNA	KAPA RNA HyperPrep	KAPA	KAPA HyperCap	AVENIO
vendor	Plation	HyperPrep	HyperPlus	Tubes	Plates	RNA-seq	HyperPrep	HyperPrep	RiboErase +Globin	Library Quant	v3	ctDNA
	Sciclone G3 NGSx	~	~	<b>~</b>	<b>~</b>	~	~	<b>~</b>	<b>~</b>	<b>~</b>	¥	~
Perkin	Sciclone G3 NGSx iQ	~	~								~	
Elmer	JANUS G3 NGS Express	~	~			~			~	~		
	Zephyr G3 NGS	~	~			~	~	~	~	<b>v</b>		
	Biomek FX <sup>p</sup> Hybrid	$\bigcirc$	$\bigcirc$			$\bigcirc$	$\bigcirc$	$\bigcirc$	$\bigcirc$	~	$\bigcirc$	$\bigcirc$
Beckman Coulter Life Sciences	Biomek i7 Hybrid	$\bigcirc$	$\bigcirc$	D	D		$\bigcirc$	$\bigcirc$	$\bigotimes$	~	$\bigcirc$	D
	Biomek i5 MC96	~	~		~							
Hamilton	NGS STAR (Span-8)	<b>v</b>	<b>v</b>	D				<b>~</b>	<b>&gt;</b>	<b>v</b>	<b>~</b>	
Tecan	Freedom EVO NGS	<b>~</b>	<b>v</b>				~	<b>~</b>	✓			
Agilent	Bravo NGS Workstation (Option A)	<b>v</b>	<b>v</b>			~				~		
ngiltill	Bravo NGS Workstation (Option B)	$\bigcirc$	$\bigcirc$			~	$\bigcirc$	~	$\bigcirc$	¥	D	
Eppendorf	epMotion 5075t	~	~			~	~	~	~	~		

### Table 4. Automated NGS library preparation workflows supported by Roche.



# AVENIO ctDNA & Tumor Tissue Analysis Kits

A versatile solution for multiple research applications

Comprising three liquid biopsy assays and three corresponding tumor tissue assays with exactly matched panels, the AVENIO NGS Oncology Assays offer a uniquely versatile solution for tumor profiling, monitoring, and concordance analysis.

### Innovative panel design and workflow

The AVENIO assays include three kits for the analysis of circulating tumor DNA (ctDNA) and three kits for analyzing tumor tissue. Each panel in the Targeted, Expanded, and Surveillance kits includes biomarkers in the U.S. National Comprehensive Cancer Network (NCCN) Guidelines.

### **Targeted Kits**

17 genes 81 kb 17 guideline-aligned genes for genomic profiling of solid tumors

### **Expanded Kits**

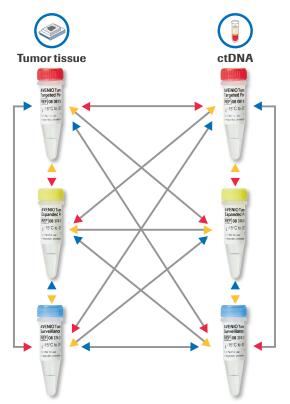
77 genes
192 kb
17 guideline-aligned genes and
60 emerging biomarkers
investigated in clinical research for
expanded profiling of solid tumors

### **Surveillance Kits**

 197 genes
 17 guideline-aligned genes, plus
 471 frequently mutated diseaseassociated regions across 197 genes; optimized for longitudinal monitoring of tumor burden in lung and colorectal cancer

### Analytical concordance

AVENIO kits are built for versatility, providing the ability to switch between tissue and plasma to support a variety of potential research applications. The analytical concordance feature in the AVENIO Oncology Analysis Software enables a simple yet detailed comparison of results across any two samples.



Exactly matched panels in corresponding ctDNA and tissue kits, as well as the inclusion of the same 17 guideline-aligned genes in all AVENIO assays, facilitate concordance analysis.

**AVENIO** assays accurately and reliably identify alterations in genes known to be somatically altered in cancer. These genes are sequenced at great depth to identify the relevant somatic alterations, including SNVs, indels, CNVs, and fusions.



# AVENIO Tumor Tissue CGP Kit

Backed by the trusted expertise and proven technology of Roche and Foundation Medicine®

# AVENIO Tumor Tissue CGP Kit

Powered by FOUNDATIONONE®

Designed to match the content of the 324-gene FoundationOne<sup>®</sup> CDx panel, the **AVENIO Tumor Tissue CGP Kit** enables labs to implement in-house comprehensive genomic profiling with an integrated end-to-end 5-day workflow solution from DNA extraction through secondary analysis with FoundationOne<sup>®</sup> Analysis Platform.



From DNA isolation to secondary analysis in 5 days



Detects 4 genomic alterations (InDels, Rearrangements, CNAs and SNVs)



Assesses 3 complex genomic signatures (TMB, MSI and LOH)



FOUNDATION

Roche

Integrated end-to-end workflow solution

# FoundationOne® Analysis Platform

Post-sequencing secondary analysis software makes it easy for labs to analyze samples to identify variants across various solid tumor types.

Details Workflow	Results QC metrics					I	
	Completed Readout						
	Mener plate type <b>Nate B</b> Dualey density	Primer well position CI Percent of cluster passing f	her.	Start data/line 21 Nov-2620 D4.45 PM PST	21 Nov-2820 5	DA AD PM PST	
	175.383011	87.91562					
Sample details Flow cell ID: H3	8 samples 874V8600 <sup>6</sup>					RNC0133152579 Analysis	
Sample ID	Test name	Primer plate type	nimer well position	Status	Comments	Pays status	
FMI#20,0091	AVENID Turner Tissue OS.	Place 8	18	Completed	Add a comment	Completed	
FMI#20_0097	AVENIO Tumor Tissue OS	Place B	2	Completed	Add a comment	Dani detarime 03-Aug-2021 04:00 PM PST End detarime	
FMI#20,0095	AVENIO Turner Tissue OG.,	Plate 8	1	Completed	Add a comment.	03-Aug 2021 10:33 PM PST	
FM1#20_0095	AVENIO Tumor Tissue OD	Pace 8	n	Completed	Add a comment.	Analysis version 0.9	
FMI#20,0092	AVENIO Tumor Tissue OS	Place 8	3	Completed	Add a comment	User name	
FMI#20,0093	AVENIO Turner Tissue OG.,	Place B 0	99	Completed	Add a comment	Output files  All results(ZP)	
FMI#20,0094	AVENIO Tumor Tissue OS	Plate 8	a	Completed	Add a comment	Downloads as one cip file	
FMI#20,0090	AVENIO Tumor Tissue 00	Plate D J	a	Completed	Add a comment.	Conserts Add a comment	
						Foundation Dre, Analysis, Platform®	

- Evidence-driven variant calling knowledge base for secondary analysis, built on unique insights from FMI's experience in profiling over 500,000+ samples
- Continuously evolving based on evidence compiled by a multidisciplinary team of cancer biologists from scientific publications, conferences, and online databases (COSMIC, dbSNP, gnomAD, 1000 Genomes)
- Web application for download of analysis output files (VCF, JSON, CSV, BAM)

Roche offers an extensive CGP portfolio that includes the AVENIO Tumor Tissue CGP Kit and navify<sup>®</sup> Mutation Profiler software for tertiary analysis.

AVENIO is a trademark of Roche.

ų,

CGP, comprehensive genomic profiling. Indels, insertion deletion. CNAs, copy number alterations. SNVs, signal nucleotide variants.

TMB, tumor mutational burden. MSI, microsatellite instability. LOH, genomic loss of heterozygosity.



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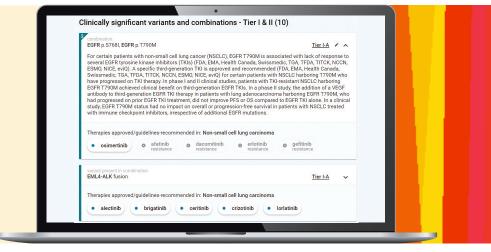
# **Bioinformatic Analysis**

Cloud-based secondary and tertiary analysis solutions

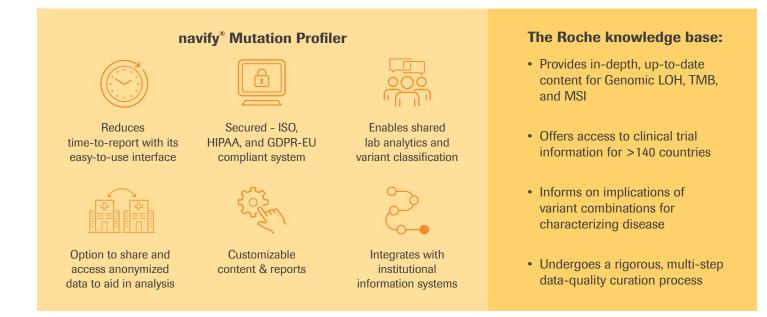
# navify<sup>®</sup> Mutation Profiler

# Cloud-based tertiary analysis for NGS assay or platform

Streamline your interpretation, annotation and reporting of oncology sequencing data with an intuitive interface and the comprehensive Roche knowledge base—with access to targeted therapies and clinical trials—for personalized medicine studies.



navity)



Roche offers an extensive CGP portfolio of flexible solutions which includes navify Mutation Profiler software and the AVENIO Tumor Tissue CGP Kit for in-house comprehensive genomic profiling.

Please visit **navify.com/products/navify-mutation-profiler** or contact your local Roche Sequencing representative for more information.



AVENIO Tumor Tissue CGP Kit is for Research Use Only. Not for use in diagnostic procedures. In the US, navify Mutation Profiler is for Research Use Only. Not for use in diagnostic procedures. AVENIO and navify are registered trademarks of Roche.

ISO, International Organization for Standardization. CGP, comprehensive genomic profiling. HIPAA, Health Insurance Portability and Accountability Act. GDPR-EU, General Data Protection Regulation European Union. LOH, genomic Loss of Heterozygosity. TMB, Tumor Mutational Burden. MSI, Microsatellite Instability

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# Validation of Sequencing Results

The Digital LightCycler<sup>®</sup> dPCR System

The Digital LightCycler<sup>®</sup> dPCR System combines sensitivity, precision, flexibility, and integration in one powerful clinical research tool. This can be used to validate variants revealed by NGS—including investigating minimal residual disease (MRD)—and much more.

This system has the potential to advance global medical knowledge by enabling researchers and physicians to push beyond the current boundaries of clinical research.



# The unique combination changing the future of digital PCR



### 3 nanowell plate configurations:

- 20,000-partition High-Sensitivity Plate (45 µl)
- 28,000-partition Universal Plate (30 µl)
- 100,000-partition High-Resolution Plate (15 μl)
- 6 advanced optical channels:
- Enable a high degree of multiplexing
- A separate channel for controls



### 5X concentrated master mixes:

 4:1 sample:master mix ratio enables more DNA or RNA sample per reaction

# The Digital LightCycler® dPCR System elevates clinical research



- Detect indels down to <0.2% allele fraction with the 20,000-partition plate.
- Detect rare mutations down to <0.1% allele fraction with the 28,000-partition plate.

### **Precision**



- Discriminate small differences between samples with the 100,000-partition plate.
- Obtain high-resolution results for absolute quantification with short turnaround times, accelerating
  publication and the development of clinically viable assays.
- Minimize the risk of amplicon contamination by virtue of the Digital LightCycler's® closed system.

Address multiple challenges at once with 6 optical channels and 3 plate configurations.Increase resolution by running a single sample on multiple lanes and combining results.

Flexibility



Choose batch size increments of between 8 and 96 samples per run.



- Integrate unique Digital LightCycler<sup>®</sup> features within a closed system to minimize contamination.
- Simplify your workflow with sample tracking through LIMS connectivity and automated data analysis.



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Contact us at: sequencing.roche.com

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