Application

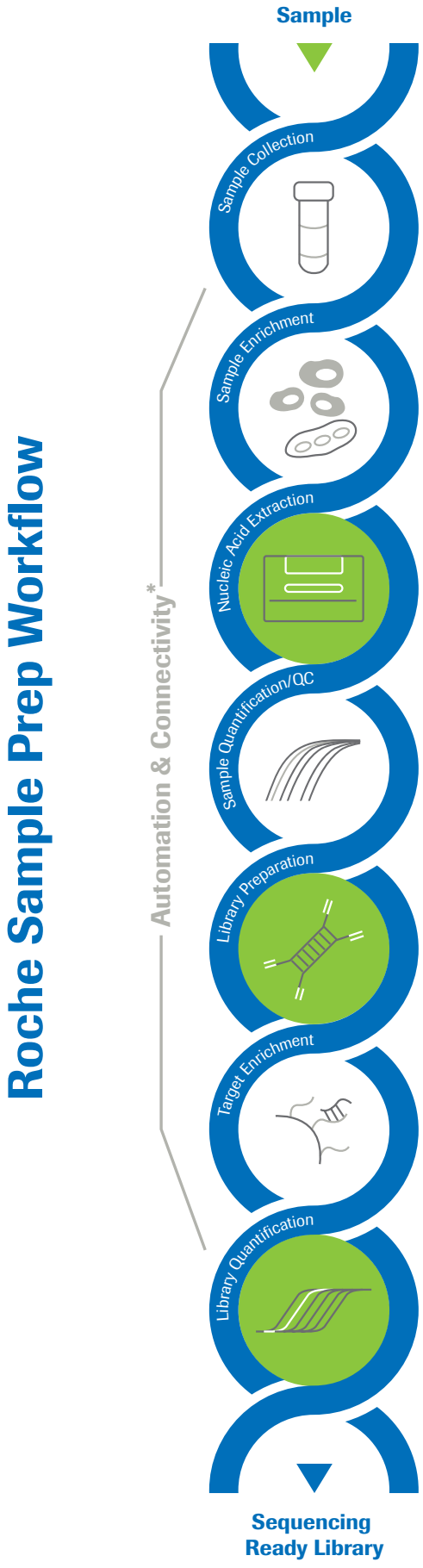
Human Whole-Genome Sequencing

Unlock the Potential of Every Sample

Application:
Human Whole-Genome Sequencing

Application Challenges

- **High-molecular weight (HMW) genomic DNA (gDNA)** is needed to make high-quality WGS libraries with 350 – 650 bp inserts for sequencing on Illumina® HiSeq® X and NovaSeq™ instruments.[1]
- Libraries that have **narrow insert size distributions**, and are **free of unwanted adapter species** help to ensure optimal clustering, mitigate the potential impact of index mis-assignment, and facilitate data analysis.
- **PCR-free workflows** are preferred to eliminate biases associated with library amplification.
- Size selection results in a significant loss of input DNA. In PCR-free workflows, **high conversion rates** are needed to produce sequencing-ready libraries at the appropriate concentration for loading HiSeq X and NovaSeq flow cells.
- PCR-free libraries can be difficult to **reliably quantify** with spectrophoretic, fluorometric or electrophoretic methods, which are unable to distinguish fragments that can be sequenced from those with only one or no adapters.
- To ensure high success rates from samples of variable quality in high-throughput pipelines, **flexibility** and **robustness** are important.



[1]https://www.illumina.com/products/by-type/sequencing-kits/library-prep-kits/truseq-dna-pcr-free.html
*Automation and Connectivity solutions are in development.
The solutions discussed here are for Research Use Only. Not for use in diagnostic procedures.
LightCycler and KAPA are for Research Use Only. Not for use in diagnostic procedures.

Solutions

NGS-qualified methods† for the **MagNA Pure 24** and **MagNA Pure 96 Nucleic Acid Extraction Systems** enable the extraction of microgram quantities of HMW genomic DNA from blood.

KAPA HyperPrep Kits offer flexible, single-tube, PCR-free library construction.

KAPA Dual-Indexed Adapters are QC-tested for NGS performance and barcode cross-contamination.

KAPA Pure Beads offer efficient, tunable reaction cleanups.

KAPA Library Quantification Kits enable accurate, qPCR-based library quantification. Compatible with Roche LightCycler® 96 and LightCycler® 480 real-time PCR systems.

†MagNA Pure 24 hgDNA ds 200 and MagNA Pure 96 DNA Blood ds SV methods

Benefits

- **Reliably extract enough human gDNA** for the construction of high-quality human WGS libraries, from as little as 200 µL EDTA-blood.
- Choose from KAPA HyperPrep protocols with **tunable** post-fragmentation or post-ligation **size selection**, to best suit your sample requirements and operational preferences.
- Achieve **very high conversion** of input DNA to adapter-ligated library, to enable PCR-free human WGS library construction from 500 ng input DNA or less.
- **Optimize your sequencing capacity** by losing fewer reads to barcode cross-contamination, index mis-assignment, short fragments and unwanted adapter species, and inaccurate normalization.
- Achieve higher throughput and consistency with **automation-friendly** KAPA HyperPrep and KAPA Library Quantification protocols.
- Enjoy greater peace of mind with **integrated support and service** for a complete human WGS workflow solution.

Unlock the Potential of Every Sample

Unlock the Potential of Every Sample

As the first step in the NGS workflow continuum, sample prep holds the key to unlocking the potential of every sample. Because NGS samples are precious, the best methods are needed to process more samples successfully, obtain more information from every sample, and optimize your sequencing resources. From sample collection to sequencing-ready libraries, Roche Sample Prep Solutions offer workflows for different sample types and sequencing applications that are **proven**, **simple** and **complete**.

App Note

KAPA HyperPrep Kits offer a flexible, high-efficiency library preparation solution for PCR-free human whole-genome sequencing. Roche Sequencing Solutions, 2018.

Webinars

Human whole-genome sequencing in the era of the Illumina® HiSeq® X and NovaSeq® platforms, featuring Bob Fulton (McDonnell Genome Institute, Washington University) and Dr. HarshaVardhan Doddapaneni (Human Genome Sequencing Center, Baylor College of Medicine). Presented by Xtalks and Roche Sequencing Solutions, 24 October 2017.

Publications

Miller, N.A. et al. A 26-hour system of highly sensitive whole genome sequencing for emergency management of genetic diseases. *Genome Medicine*. 2015;7,100. doi:10.1186/s13073-015-0221-8

Hiranuma, N. et al. Cis-compound mutations are prevalent in triple negative breast cancer and can drive tumor progression. *bioRxiv*. November 2016. doi:10.1101/085316

Srivastava, A. Genomes of the Mouse Collaborative Cross. *Genetics*. 2017;206, 537. doi:10.1534/genetics.116.198838

Ordering information

Roche Cat. No.	Kapa Code	Description	Pack Size
07290519001	N/A	MagNA Pure 24 Instrument	1 instrument
06541089001	N/A	MagNA Pure 96 Instrument	1 instrument
07658036001	N/A	MagNA Pure 24 Total NA Isolation Kit	Up to 96 extractions
06543588001	N/A	MagNA Pure 96 DNA and Viral NA Small Volume Kit	576 isolations
07962355001	KK8503	KAPA HyperPrep Kit, PCR-free	24 reactions
07962371001	KK8505	KAPA HyperPrep Kit, PCR-free	96 reactions
08278555702	KK8722	KAPA Dual-Indexed Adapter Kit (15 µM)	96 x 20 µL
07983280001	KK8001	KAPA Pure Beads	30 mL
07983298001	KK8002	KAPA Pure Beads	60 mL
07960140001	KK4824	KAPA Library Quantification Kit (Illumina/Universal)	500 reactions
07960298001	KK4854	KAPA Library Quantification Kit (Illumina/LC480)	500 reactions

Published by:

Roche Sequencing Solutions, Inc.
4300 Hacienda Drive
Pleasanton, CA 94588

sequencing.roche.com

SEQ100185 06/18