

TruPath™ Genome performance with varying sample sources and extraction methods

Compatible with a range of sample types, including blood, cells, saliva, dried blood spots, and buccal swabs

High-quality results from a range of sample qualities, including DNA from standard and high molecular-weight extraction kits

Ideally suited for human genome phasing studies with various sample types and extraction methods

Introduction

Short-read next-generation sequencing (NGS) offers a flexible, reliable method for performing high-accuracy whole-genome sequencing (WGS). However, a small portion of the human genome remains difficult to map, eg, regions with high sequence homology, repetitive sequences, or structural complexity. Long-read sequencing methods can help resolve these regions but are hindered by complex workflows and accuracy challenges that can lead to variable results.¹⁻⁴

Illumina TruPath Genome revolutionizes the short-read NGS workflow, providing comprehensive WGS with unprecedented simplicity.⁵ Powered by **proximity mapped read technology**, TruPath Genome uses a highly streamlined workflow that goes from purified DNA to loading the sequencing system in ~10 minutes (**Figure 1**). Sequencing occurs on the NovaSeq™ X Plus System with DRAGEN™ secondary analysis. The resulting high-accuracy short-read data is combined with nanowell proximity information from DNA templates on the flow cell. This proximity information allows labs to

generate long-distance genomic insights for sequences separated by up to millions of bases. Integrating these long-distance genomic insights with the strengths of short-read sequencing allows TruPath Genome to make comprehensive genomes accessible.

TruPath Genome supports purified double-stranded DNA (dsDNA) extracted using a commercial kit appropriate for the sample type.* This technical note evaluates the compatibility of TruPath Genome with different DNA extraction methods and correspondent sample types, including whole blood, cells, saliva, dried blood spot (DBS) cards, and buccal swabs. Results demonstrate the highly robust capabilities of TruPath Genome to resolve previously difficult-to-map regions of the genome and perform comprehensive variant detection with a breadth of sample types and extraction methods.

* DNA isolated from formalin-fixed paraffin-embedded (FFPE) and cell-free DNA (cfDNA) samples are not supported.

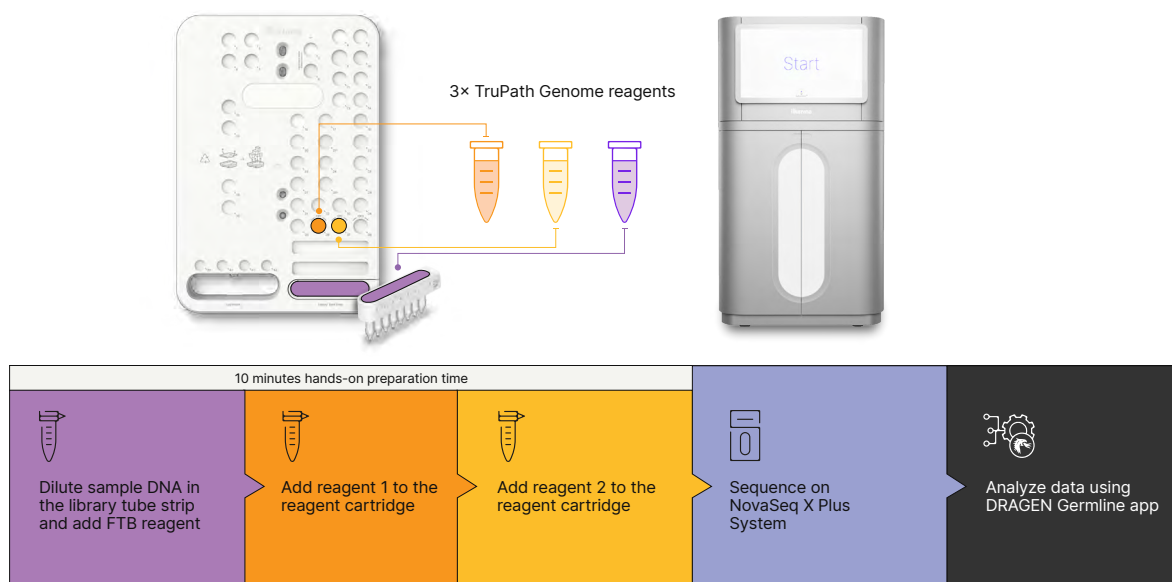


Figure 1: Streamlined TruPath Genome workflow

Purified DNA is added directly to the library tube strip along with FTB (Flow Cell Tagment Buffer) reagent and water to the desired concentration. The tube strip and remaining TruPath reagents are added to the sequencing cartridge. Next, the cartridge and flow cell are loaded into the NovaSeq X Plus System and the run is started. Hands-on time to set up the assay is 10 minutes.

Methods

Samples and DNA extraction

A range of sample types, including whole blood, cells, saliva, DBS, and buccal swabs were used to assess TruPath Genome performance. A minimum of three donors were used for each sample type. DNA extraction kits were selected based on the following criteria:

- The isolated nucleic acid should be suitable for downstream library prep and sequencing
- A range of purification methods were included (eg, spin-column, magnetic beads, alcohol precipitation) where possible, depending on sample type
- High molecular weight (HMW) and standard DNA extraction methods were included for blood and cell samples
- Commercially established extraction methods widely used in routine laboratory workflows

Whole blood

Blood samples stored in K₂EDTA or sodium heparin from healthy donors were purchased from Research Donors (London, UK). The cohort consisted of six healthy donors. Six extraction kits were evaluated for blood samples stored in K₂EDTA, and three extraction kits were evaluated for blood samples stored in sodium heparin (Table 1). HMW and standard DNA extraction methods were included. DNA was extracted following the protocol for mammalian whole blood. To obtain high-quality DNA, samples can be stored for up to three days at 2°C to 8°C. For three of the six extraction methods tested, an additional extraction was performed for blood stored for seven days at 4°C.

Mammalian cell pellets

Cultures of lymphoblastoid or fibroblast cell lines were ordered from the Coriell Institute for Medical Research. Cultures were harvested by centrifugation and washed twice with phosphate buffered saline (PBS). Cell counts were determined using a cell counter, and 1×10^6 – 2×10^6 cell pellets were prepared either for immediate extraction (referred to as fresh in Table 2) or frozen at -80°C.

Table 1: DNA extraction methods used for whole blood samples

No. of samples	Sample storage	DNA extraction kit	Method	HMW specific	Comments
3	< 3 days at 4°C or 7 days at 4°C	Monarch HMW DNA Extraction Kit for Cells & Blood (New England Biolabs, Catalog no. T3050L) ^a	Glass beads	Yes	Shaking incubation speed for lysis was performed at 1200 RPM at 56°C for 10 min
3	< 3 days at 4°C	Wizard HMW DNA Extraction Kit (Promega, Catalog no. A2920)	Alcohol precipitation	Yes	No changes in protocol
3	< 3 days at 4°C or 7 days at 4°C	MagAttract HMW DNA Kit (QIAGEN, Catalog no. 67563)	Magnetic beads	Yes	No changes in protocol
3	< 3 days at 4°C or 7 days at 4°C	QIAamp DNA Blood Mini Kit (QIAGEN, Catalog no. 51104) ^a	Spin column	No	No changes in protocol
3	< 3 days at 4°C	Mag-Bind Blood & Tissue DNA HDQ 96 Kit (Omega Bio-tek, Catalog no. M6399)	Magnetic beads	No	No changes in protocol
3	< 3 days at 4°C	Chemagic Prepito DNA Universal Kit (Revvity, Catalog no. CMG-2034) ^a	Magnetic beads	No	No changes in protocol; protocol was run on the Chemagic Prepito instrument (Revvity, Catalog no. 2022-0030)

a. Extraction kit was used to evaluate blood samples stored in sodium heparin as well as K₂EDTA. Blood samples stored in sodium heparin were stored for < 3 days at 4°C. HMW, high molecular weight; RPM, revolutions per minute.

Two extraction kits were evaluated (Table 2). DNA was extracted following the protocol for mammalian cells. In addition, HMW and standard DNA samples from the Coriell Institute for Medical Research were also evaluated; these included reference samples from the Genome in a Bottle Consortium (GIAB): HG001, HG002, HG003, HG004, HG005, HG006, and HG007. For the GM24385/HG002 cell line, additional extraction kits were evaluated to assess DNA quality (Table 3).

Dried blood spots

The cohort consisted of three healthy donors. DBS were prepared using 50 µl of < 3 days old K₂EDTA-whole blood in Whatman 903 Protein Saver Cards. Limited testing on Revvity 226 Spot Saver Cards indicates these are also compatible with TruPath Genome (data not shown). The DBS cards were kept at room temperature (RT) for at least three hours to dry. Dried cards were stored inside a sealed bag containing desiccant packs at RT. Performance with TruPath Genome was evaluated

Table 2: DNA extraction methods used for mammalian cells

No. of samples	Sample storage	DNA source/extraction kit	Method	HMW specific	Comments
35	Fresh or frozen at -80°C for < 1 year	Monarch HMW DNA Extraction Kit for Cells & Blood (New England Biolabs, Catalog no. T3050L)	Glass beads	Yes	Shaking incubation speed for lysis was performed at 1200 RPM at 56°C for 10 min
26	Fresh or frozen at -80°C for < 1 year	QIAamp DNA Blood Mini Kit (QIAGEN, Catalog no. 51104)	Spin column	No	No changes in protocol
9	Unknown	Coriell HMW DNA ^a	N/A	Yes	N/A
7	Unknown	Coriell Standard DNA ^a	N/A	No	N/A
a. Additional sample information is provided by the supplier. HMW, high molecular weight; RPM, revolutions per minute.					

Table 3: DNA extraction methods used for GM24385 mammalian cells

Sample storage	DNA extraction kit	Method	HMW specific	Comments
Fresh or frozen at -80°C for < 1 year	Monarch HMW DNA Extraction Kit for Cells & Blood (New England Biolabs, Catalog no. T3050L)	Glass beads	Yes	Shaking incubation speed for lysis was performed at 1200 RPM at 56°C for 10 min
	Wizard HMW DNA Extractio Kit (Promega, Catalog no. A2920)	Alcohol precipitation	Yes	No changes in protocol
	QIAamp DNA Blood Mini Kit (QIAGEN, Catalog no. 51104)	Spin column	No	No changes in protocol
	Mag-Bind Blood & Tissue DNA HDQ 96 Kit (Omega Bio-tek, Catalog no. M6399)	Magnetic beads	No	No changes in protocol
	Chemagic Prepito DNA Universal Kit (Revvity, Catalog no. CMG-2034)	Magnetic beads	No	Cells resuspended in 250 µl PBS; blood protocol was run on the Chemagic Prepito instrument (Revvity, Catalog no. 2022-0030)
Unknown	Coriell HMW DNA ^a	N/A	Yes	N/A
Unknown	Coriell Standard DNA ^a	N/A	No	N/A
a. Additional sample information is provided by the supplier. HMW, high molecular weight; PBS, phosphate buffered saline; RPM, revolutions per minute.				

for DBS cards stored for either one month or one year. Four extraction kits were evaluated (Table 4). DNA was extracted following the protocol for DBS cards, using 6 × 3 mm² punches per extraction.

Saliva

Saliva samples from healthy donors were purchased from Research Donors (London, UK). These were collected using two devices: Genefix Saliva DNA & RNA Collector (Isohelix, Catalog no. GFX-02) and Oragene Dx Saliva Collection Kit (DNA Genotek, Catalog no. OGD-600) according to kit instructions. Samples were used within one year of collection. Cohorts consisted of three healthy

donors. Three extraction kits were evaluated for samples collected with the Oragene Dx Saliva Collection Kit and two extraction kits were evaluated for samples collected with the Genefix Saliva DNA & RNA Collector (Table 5).

Buccal swabs

Buccal swab samples from three healthy donors were purchased from Research Donors (London, UK). These were collected using the ORACollect•DNA self-collection kit (DNA Genotek, Catalog no. OCR-100), according to manufacturer instructions. Samples were used within 60 days of collection. Two extraction kits were evaluated for the collected samples (Table 6).

Table 4: DNA extraction methods used for DBS samples

No. of samples	Sample storage	DNA extraction kit	Method	Comments
3	< 1 month at RT or ~ 1 year at RT	Chemagic Prepito DNA Universal Kit (Revvity, Catalog no. CMG-2034)	Magnetic beads	Prelysis at 56°C for 3 hr at 800 RPM; DBS protocol was run on the Chemagic Prepito instrument (Revvity, Catalog no. 2022-0030)
3	< 1 month at RT or ~ 1 year at RT	SparQ Lysis Kit (Quantabio, Catalog no. 95220-096)	Magnetic beads	Supplemental protocol provided by supplier; heat inactivation post-lysis was performed at 70°C for 10 min
3	< 1 month at RT or ~ 1 year at RT	ReliaPrep FFPE gDNA Miniprep System (Promega, Catalog no. A2351)	Spin column	Supplemental protocol provided by supplier
3	< 1 month at RT or ~ 1 year at RT	MagMax DNA Multi-Sample Ultra 2.0 Kit (Thermo Fisher Scientific, Catalog no. A36570)	Magnetic beads	Supplemental protocol provided by supplier; prelysis shaking for 1 hr at 65°C at 600 RPM; protocol was run on Kingfisher Flex

RPM, revolutions per minute; DBS, dried blood spot.

Table 5: DNA extraction methods used for saliva samples

No. of samples	Collection device	DNA extraction kit	Method	Comments
3	Oragene Dx Saliva Collection Kit ^a	prepIT•L2P DNA extraction reagent (DNA Genotek, Catalog no. PT-L2P)	Alcohol precipitation	500 µl input
3		Chemagic Prepito DNA Universal Kit (Revvity, Catalog no. CMG-2034)	Magnetic beads	250 µl input; blood protocol was run on the Chemagic Prepito instrument (Revvity, Catalog no. 2022-0030)
3		MagMax Saliva gDNA Isolation Kit (Thermo Fisher Scientific, Catalog no. A39060)	Magnetic beads	500 µl input
3	Genefix Saliva DNA & RNA Collector ^b	GeneFix Saliva-Prep 2 DNA Isolation Kit (Isohelix, Catalog no. GSPN)	Alcohol precipitation	500 µl input
3		Chemagic Prepito DNA Universal Kit (Revvity, Catalog no. CMG-2034)	Magnetic beads	250 µl input; blood protocol was run on the Chemagic Prepito instrument (Revvity, Catalog no. 2022-0030)

a. Samples were incubated at 50°C for three hours before DNA isolation, according to the manufacturer's instructions.

b. Samples were heated at 60°C for 1 hour with Proteinase K solution before DNA isolation, according to the manufacturer's instructions.

Table 6: DNA extraction methods used for buccal swabs samples

No. of samples	Collection device	DNA extraction kit	Method	Comments
3	OCR-100 ^a	prepIT•L2P DNA extraction reagent (DNAGenotek, Catalog no. PT-L2P)	Alcohol precipitation	250 µl input; saliva protocol followed
3		Chemagic Prepito DNA Universal Kit (Revvity, Catalog no. CMG-2034)	Magnetic beads	250 µl input; blood protocol was run on the Chemagic Prepito instrument (Revvity, Catalog no. 2022-0030)
a. Samples were incubated at 50°C for three hours before DNA isolation, according to the manufacturer's instructions.				

DNA quality assessment

Extracted DNA quantity was measured using the Qubit dsDNA High Sensitivity Assay on the Qubit 4 Fluorometer (Thermo Fisher Scientific, Catalog no. Q33238). DNA quality was assessed with one of the following methods:

- Genomic DNA ScreenTape assay (Agilent, Catalog no. 5067-5366) on the 4200 TapeStation System (Agilent, Catalog no. G2991BA); the region analysis tool was used to assess proportions greater than 60 kb and 10 kb
- A Femto Pulse gDNA 165 kb kit (Agilent, Catalog no. FP-1002-0275); the Genomic Quality Number (GQN) was determined using the 10 kb and 60 kb threshold (FEMTO Pulse System, Agilent, Catalog no. M5330AA)
- $Abs_{260/280}$ was measured using the NanoDrop spectrophotometer (Thermo Fisher Scientific, Catalog no. ND-ONE-W)

Run setup and sequencing

DNA was added to a sample library tube strip along with TruPath Genome reagents (Illumina, Catalog no. 20157406) according to the manufacturer's instructions. The standard input amount was 350 ng DNA; however, DBS samples used 175–350 ng.⁶ The minimum DNA input was 175 ng. The library tube strip, TruPath Genome reagents, and a NovaSeq X C8 flow cell were loaded on to the NovaSeq X Plus System (Illumina, Catalog no. 20084804) for sequencing according to the user manual.

Analysis

After sequencing, the DRAGEN Germline 4.5.2 pipeline was used to combine short-read sequencing data with nanowell proximity information. The GRCh38 reference genome was used for variant calling from phased reads.

Results

DNA quality from various extraction kits

Various extraction kits and methods were evaluated to determine the resulting DNA yield and integrity. For blood and cell line samples, both HMW and standard DNA quality extraction methods were used. $Abs_{260/280}$ ratios were found to be in the acceptable range of 1.8–2.0 for all extraction kits and samples (data not shown).

TapeStation analysis

DNA quality can be assessed by the gDNA ScreenTape assay, which quantifies the fragment percentage above a certain threshold. For the best performing proximity data, TruPath Genome recommends that at least 70% of the DNA fragments are above 10 kb and at least 40% of the DNA fragments are above 60 kb thresholds. At a minimum, 50% of the DNA fragments should be above 10 kb. In this study, the highest quality was observed for DNA extracted from blood and cell pellet samples using HMW DNA kits (Figure 2 and Figure 3). All extraction methods, except for one, met the minimum 10 kb threshold (Figure 2 and Figure 3).

Femto Pulse analysis

For a subset of samples, DNA quality was also assessed using the Femto Pulse System using the gDNA 165 kb assay. As with the ScreenTape assay, the percentage of fragments higher than 10 kb and 60 kb was assessed by calculating the GQN10kb and GQN60kb. This method is more accurate compared to the ScreenTape assay as it enables analysis of DNA size up to 165 kb. Results follow the same trend observed for the ScreenTape assay, where the highest quality was observed for DNA extracted from blood and cell pellet samples using HMW DNA kits (Figure 4). The genomic DNA from these samples has a GQN of at least 7.0 or higher at 10 kb

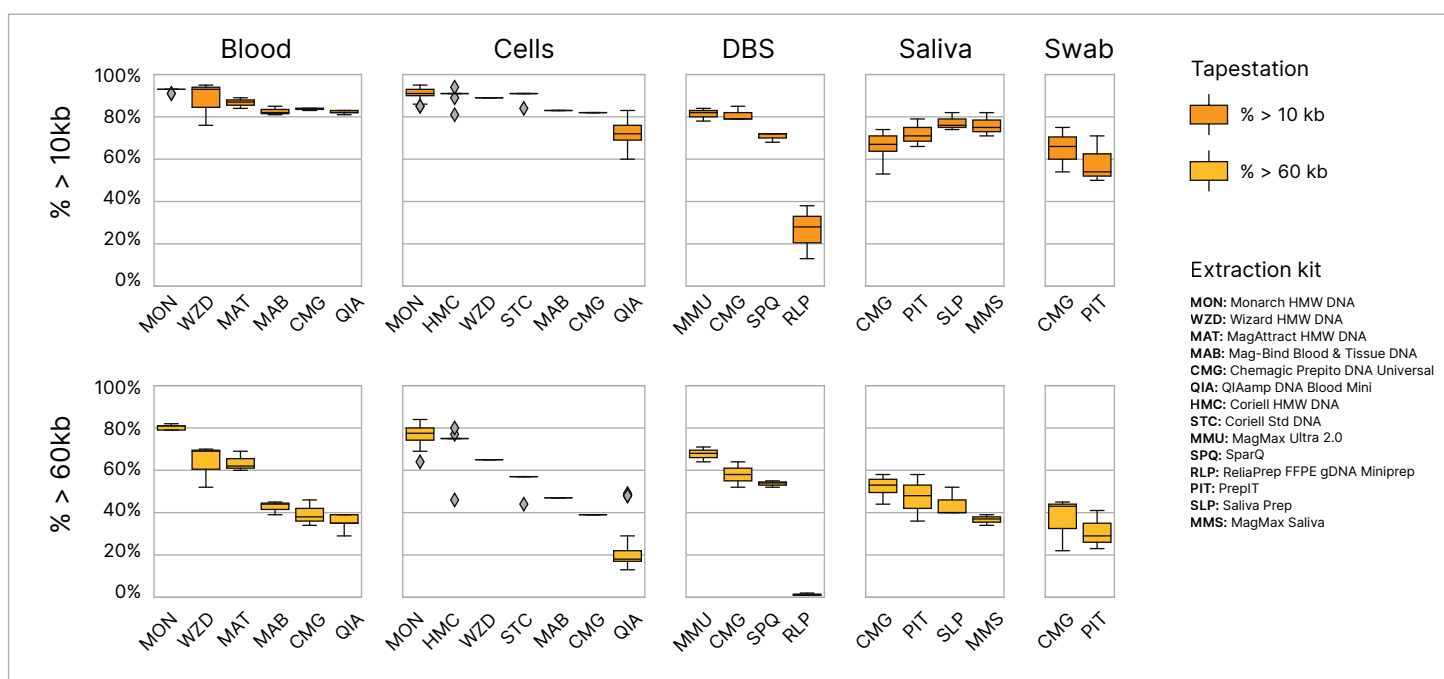


Figure 2: TapeStation analysis of DNA quality by sample type

A range of DNA quality is observed across different sample types and extraction kits. DNA was extracted from blood and cell samples using both HMW and standard commercially available kits. DNA was extracted from DBS, saliva, and buccal swabs using standard commercially available kits. Analysis of DNA quality was performed using gDNA ScreenTape assay on the Agilent 4200 TapeStation by assessing the fragment percent greater than 10 kb and 60 kb. DBS, dried blood spots.

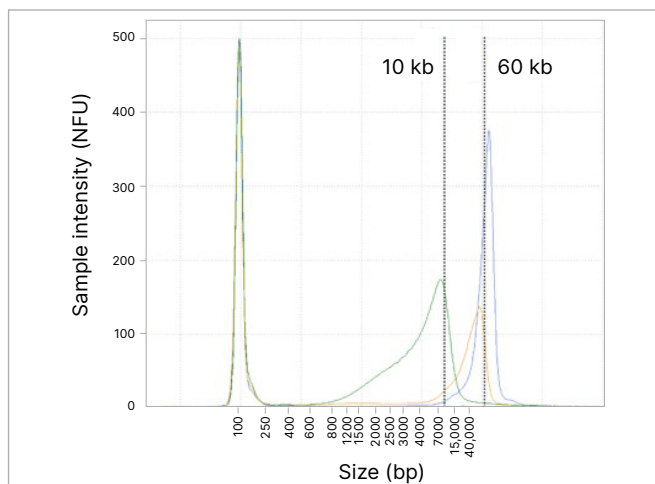


Figure 3: ScreenTape curves by sample type

Analysis of DNA quality using genomic DNA ScreenTape assay on the Agilent 4200 TapeStation for cells reveals varying quality by sample type, including whole blood extracted with a HMW DNA kit (blue), buccal swabs (orange), and DBS (green) samples, as measured by normalized fluorescence units (NFU).

and a GQN of 4.0 or higher at 60 kb. Similar results are observed among the TapeStation and Femto Pulse assays for the remaining data, where there is only one extraction method where the percentage of fragments above the 10 kb threshold is lower than 50%.

The percentages of fragments > 10 kb were concordant between the two methods used for analysis, particularly for blood and cell samples, indicating consistent detection of low-range HMW DNA. The gDNA ScreenTape assay slightly underestimates the percentage of fragments higher than 10 kb by about 5–15%, depending on the sample type and extraction kit (data not shown). However, for fragments > 60 kb, the values diverged more substantially, with the Femto Pulse System providing values that are considered more accurate for very large fragments due to its higher resolution. Nevertheless, both methods report the same trend and even though the Femto Pulse System offers greater accuracy for HMW DNA, the TapeStation remains advantageous for routine use due to its accessibility and widespread availability in most laboratories.

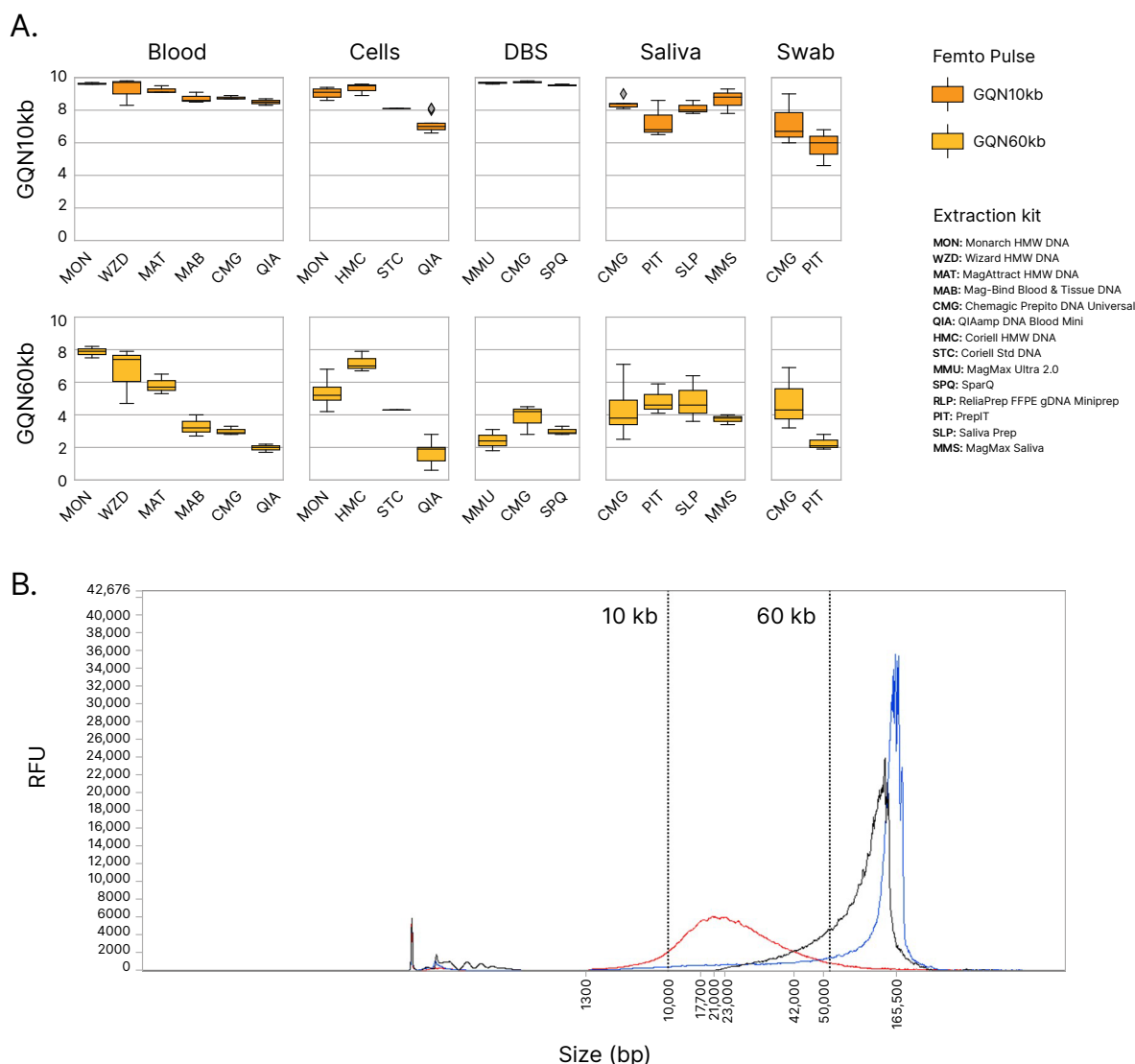


Figure 4: Femto Pulse analysis of DNA quality by sample type

(A) A range of DNA quality is observed across different sample types and extraction kits. DNA was extracted from blood and cell samples using both HMW and standard commercially available kits. DNA was extracted from DBS, saliva, and buccal swabs using standard commercially available kits. Analysis of DNA quality was performed using the Genomic DNA 165 kb kit on the Femto Pulse System by determining the GQN10kb and GQN60kb. (B) Analysis of DNA quality on FEMTO Pulse system reveals varying quality by sample type, including cells (blue, HMW), buccal swabs (black), and DBS (red), as measured by relative fluorescence units (RFU).

TruPath Genome delivers high-quality results from samples of variable quality

Standard WGS metrics, including autosomal coverage and base call accuracy, with TruPath Genome were unaffected by DNA quality and robust performance was observed for all sample types. The average autosomal coverage was ~64x and the average Q30 was 92% (Figure 5A). Slightly lower coverage was observed for

some of the saliva and swab samples, most likely due to the natural presence of bacteria, which reduced human genome mapped read percentage. The estimated bacterial content in these sample types is ~10%.⁷ This is highlighted when separating the results by sample donor (Figure 5B), where donor sample three shows lower coverage, possibly due to higher bacterial content in this sample.

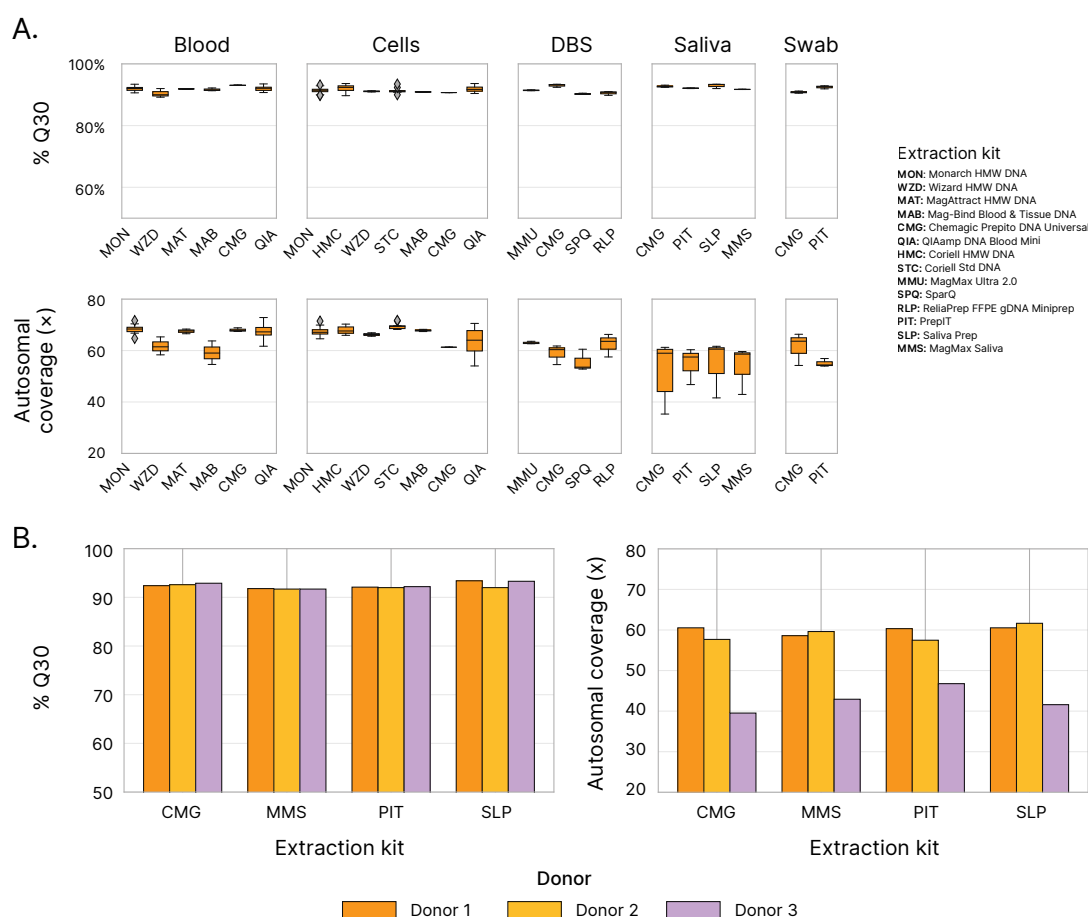


Figure 5: TruPath Genome genome coverage and quality performance is highly robust with variable sample types

(A) Autosomal coverage and average percent Q30 are not impacted by DNA quality across different sample types. Strong performance for coverage and quality scores was observed with all samples, regardless of DNA purification method. (B) Autosomal coverage by donor for saliva samples. Donor 3 shows the lowest coverage, possibly due to higher bacterial contamination.

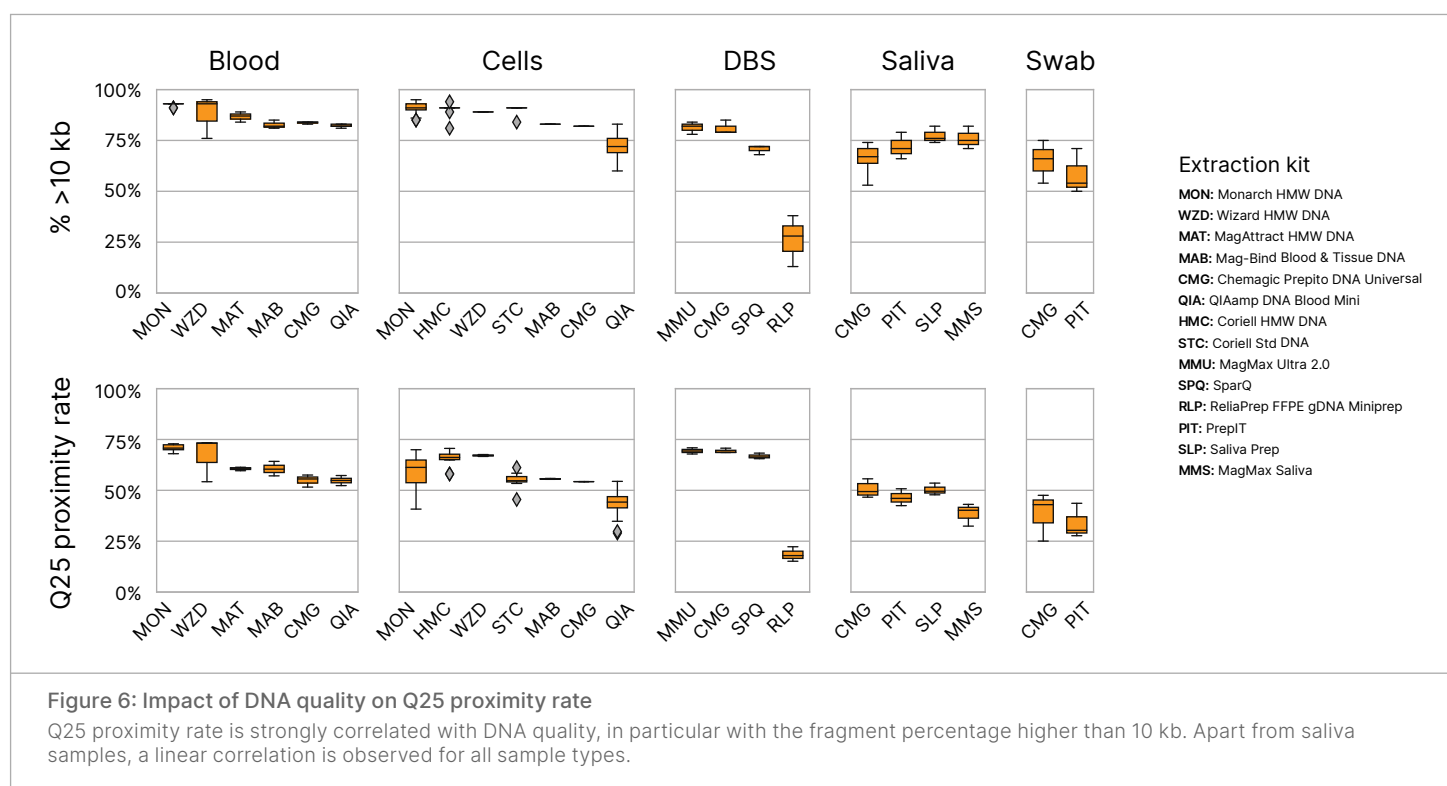
DNA sample quality was strongly associated with TruPath Genome proximity metrics (Figure 6). For most sample types, the percentage of DNA fragments over 10 kb was a strong predictor of the proximity rate performance, with a higher fragment percentage over 10 kb producing a higher Q25 proximity rate (ie, the percentage of reads that have at least one other read in close proximity with a quality score above Q25[†]).

TruPath Genome supports phased sequencing

TruPath Genome enables the study of full-length DNA templates, making it well suited for human genome phasing studies. The presence of larger DNA fragments is a strong predictor of the phasing performance, ie, the higher the DNA fragment percentage over 60 kb, the larger the size of the phasing block NG50[‡] and the higher the template length. As with the proximity rate,

[†] The proximity quality score is the Phred-scaled likelihood that two reads from the same region of the genome landed within the same flow cell neighborhood by chance as calculated by the DRAGEN proximity model. A higher proximity quality score indicates higher confidence that two reads that are nearby in the genome and on the flow cell originated from the same input DNA molecule.

[‡] The phasing block NG50 metric provides insight into the distribution of phasing block size, depicting the value threshold of 50% relative to the true length of the genome aligned contig. A greater NG50 value indicates that larger, more contiguous regions of the genome can be readily phased.



the same correlation is observed when plotting the GQN60kb vs phasing block NG50 and template length (data not shown). Results with TruPath Genome show the correlation between template length and phasing block NG50 and percentage of fragments higher than 60 kb per sample type and extraction kit, making it well suited for human genome phasing studies with various sample types and extraction methods (Figure 7).

TruPath Genome performs best with high-quality, fresh samples

To assess the impact of sample age on TruPath Genome performance, DNA was extracted from blood and DBS samples at two specific time points. For blood samples, DNA was extracted within three days of blood collection and after seven days. Samples were stored at 4°C. For DBS samples, fresh blood (less than three days old) was collected in Whatman 903 Protein Saver Cards and stored either for one month or one year at room temperature.

For both sample types, the size of the phasing block NG50 changed in relation to the percentage of DNA fragments over 60 kb (Figure 8). Both metrics were lower for the aged samples compared with fresh samples. Interestingly, for blood samples, over 40% of fragments > 60 kb were obtained even for samples where DNA was isolated after one week when DNA was extracted using an HMW method (eg, Monarch HMW DNA, MagAttract). The same correlation is observed for other proximity metrics, eg, Q25 proximity rate and template length (data not shown).

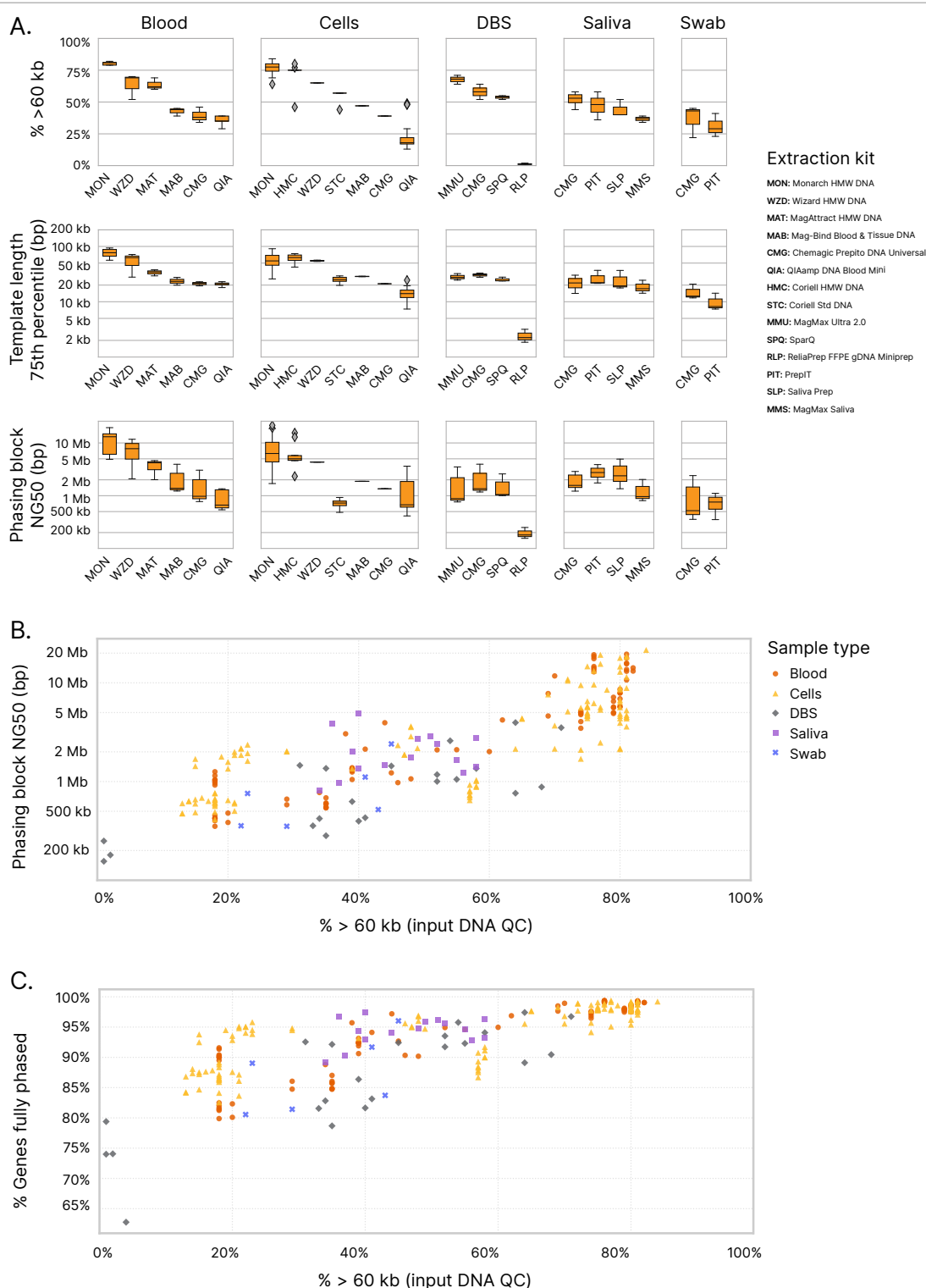


Figure 7: TruPath Genome phasing performance is highly robust across different sample types

DNA was extracted from blood and cells using HMW and standard methods; DNA was extracted from DBS, saliva, and buccal swabs using standard methods. DNA intactness was measured on the Agilent 4200 TapeStation with a regional analysis and fragment percent > 60 kb binned along the X-axis. (A) Template length on the Y-axis represents the 75th percentile template molecule size. Increased DNA quality, with larger percentage > 60 Kb, correlates with increased template length and phasing block NG50. Input DNA with a larger percentage > 60 Kb correlated with (B) phasing block NG50 and (C) % genes fully phased. DBS, dried blood spot; HMW, high molecular weight; QC, quality control.

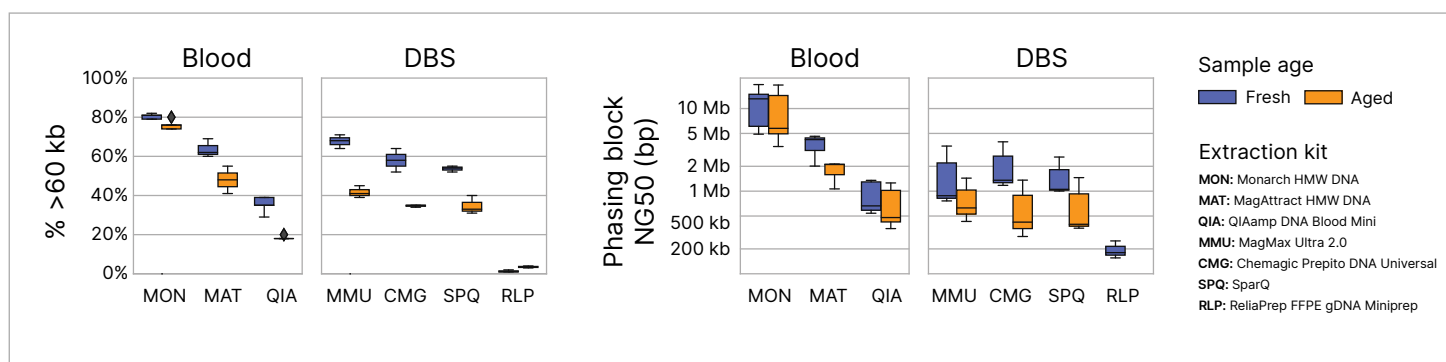


Figure 8: Impact of blood sample storage on DNA quality and phasing block NG50

Blood samples were stored at 4°C for < 3 days (fresh) or 7 days (aged). DBS samples were stored at room temperature for < 30 days (fresh) or ~1 year (aged). DNA was extracted from blood using HMW and standard methods. DNA was extracted from DBS samples using standard methods. Impact of primary sample age on DNA quality (percent > 60 kb) and phasing block NG50. Percent > 60 kb is measured using genomic DNA in the ScreenTape assay on the Agilent 4200 TapeStation. For both sample types, the percentage of fragments higher than 60 kb and the phasing block NG50 size are lower for aged samples (orange) when compared to fresh samples (blue).

Impact of sodium heparin on TruPath Genome performance with whole blood samples

Whole blood samples can be stored in sodium heparin; however, heparin is known to be a common inhibitor of downstream molecular reactions.⁸ Therefore, TruPath Genome performance on whole blood samples stored in sodium heparin was evaluated. The DNA quality of heparin blood samples was similar to that of K₂EDTA blood samples (data not shown). A slight inhibition of TruPath genome performance was observed, reflected in an increased percentage of duplicate reads, resulting in decreased coverage (Figure 9). Even so, proximity metrics, including Q25 proximity rate, size of NG50 phasing block, and the percentage of fully phased genes, are comparable between heparin and K₂EDTA blood samples (Figure 10). Observed differences in performance between samples extracted with the Monarch HMW DNA kit and QIAamp DNA Blood may be because a spin column-based purification method is more likely to remove contaminants.

Interestingly, when performing the TruPath Genome workflow using Prepito-extracted DNA, the sequencing failed. For that reason, the extracted DNA was subjected to an additional clean-up using the Genomic DNA Clean & Concentrator-10 (Zymo Research, Catalog no. D1010), following the manufacturer's instructions. A slight decrease in quality was observed (data not shown). Nevertheless, after this additional step, it was possible to obtain sequencing data comparable to Monarch HMW DNA and QIAamp DNA Blood. Despite successful sequencing following an additional DNA clean-up step for selected extraction methods, K₂EDTA remains the recommended blood collection tube because impacts on performance, including increased duplicate rates, were still observed. Furthermore, the effectiveness of the clean-up approach may vary across extraction methods, and these findings are based on a limited sample set.

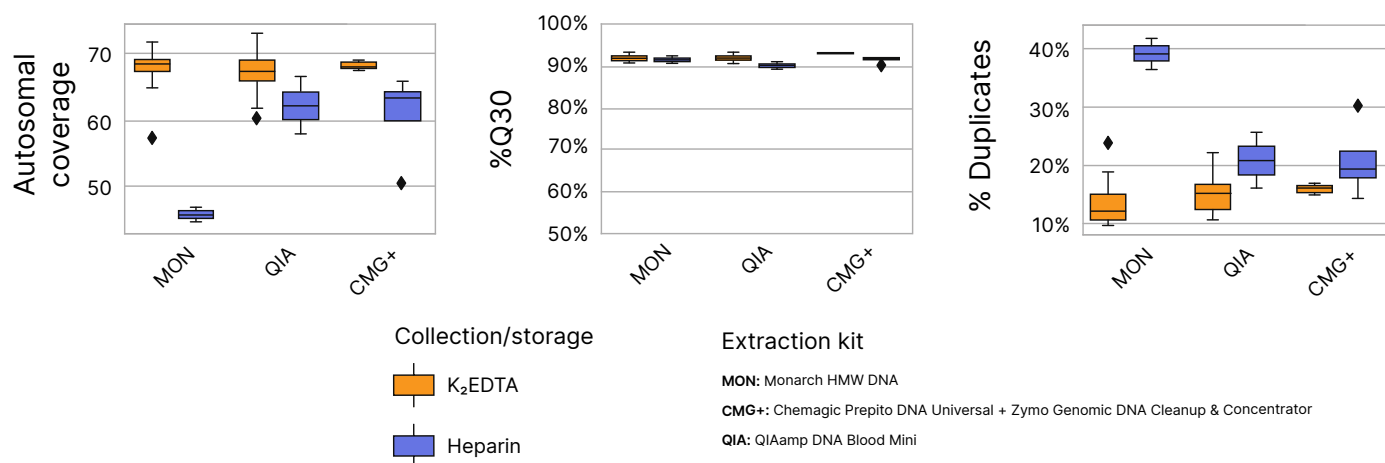


Figure 9: Slight inhibition in TruPath Genome performance with heparin blood samples

A slight drop in coverage is observed due to the increased duplicate percentage, especially for Monarch extracted DNA. Chemagic Prepito extraction includes an additional clean-up for sodium heparin blood samples.

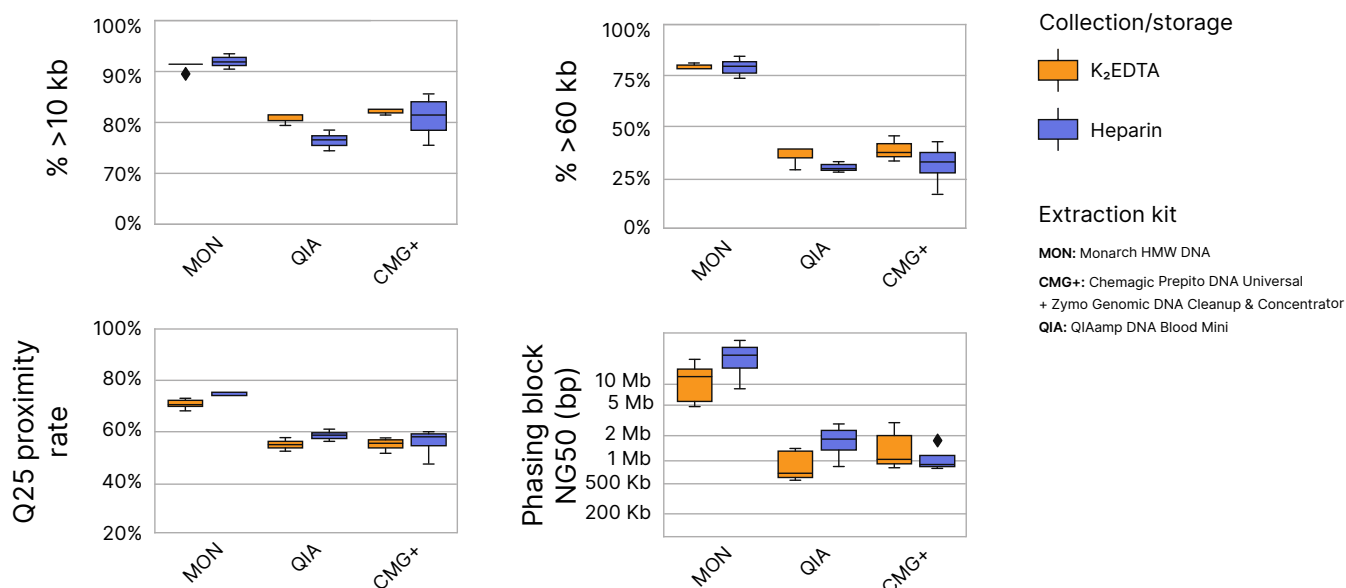


Figure 10: TruPath Genome phasing performance is comparable with K₂EDTA and heparin blood samples

A correlation was observed between Q25 proximity rate and percentage of fragments higher than 10kb and NG50 phasing block and the percentage of fragments higher than 60kb, respectively. Chemagic Prepito extraction includes an additional clean-up for sodium heparin blood samples.

Summary

The compatibility of different sample types and extraction methods with TruPath Genome was evaluated. DNA quality is highly dependent on the sample and on the extraction method. To obtain the best proximity data possible, it is recommended to have DNA showing at least 70% of fragments larger than 10 kb and 40% of fragments larger than 60 kb. At a minimum, at least 50% of the fragments should be higher than 10kb. Together, these results demonstrate the high-quality, robust performance achieved with TruPath Genome from different sample sources, including blood, cells, DBS, saliva, and swabs.

Learn more →

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