# Enhanced Detection of Structural Variants, VNTRs, and Haplotype Phasing with SBX Simplex Longer Sequencing (SBX-SL)

**Chen Zhao,**[2] Aaron Jacobs,[1] Alberto Gatto,[2] Alex Lehmann,[1] Alex Mitchell,[1] Chad Rosevear,[1] Cynthia Cech,[1] Daniel Zinder,[2] Dilmi Perera,[2] Donald Pacaba,[1] Erfan Sayyari,[2] Grant Kingsley,[1] Jagadeeswaran Chandrasekar,[1] Janie Johnson,[1] John Mannion,[2] Jordan Eizenga,[2] Kendall Berg,[1] Kevin Smith,[1] Lacey McGee,[1] Mahdi Golkaram,[2] Majid Babazadeh,[1] Marc Prindle,[1] Melanie Kirsche,[2] Melud Nabavi,[1] Mitchell Wolfin,[1] Robert McRuer,[1] Salka Barrett,[1] Sam Salari,[2] Scott Miller,[1] Svetlana Kritzer,[1] Thomas Reid,[1] Upneet Bala,[1] Yanli Hou,[2] Mark Kokoris[1]

[1]Roche Sequencing Solutions, Inc, Seattle, WA, USA; [2]Roche Sequencing Solutions, Inc, Santa Clara, CA, USA; [3]Roche Diagnostics GmbH, Penzberg, Germany

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## Introduction

Sequencing By Expansion (SBX) is a high-throughput, flexible, and rapid-turnaround sequencing technology.[1] While SBX duplex sequencing (SBX-D) offers high accuracy for detecting small germline variants such as SNVs and INDELs, its ability to resolve larger genomic features—such as structural variants (SVs), tandem repeats (TRs), and long-range haplotypes—is limited. Here, we present SBX Simplex Longer sequencing (SBX-SL), an extension of the SBX platform that significantly enhances the detection of SVs and improves haplotype phasing in whole-genome sequencing (WGS) workflows. SBX data were processed through SBX-optimized open source (XOOS) variant callers.[2]

## Introduction to SBX-SL

SBX Simplex Longer (SBX-SL) is a library prep and sequencing protocol that is able to achieve longer read lengths than the standard SBX duplex (SBX-D) workflow. It includes adapter ligation, linear amplification, Xpandomer synthesis, and sequencing

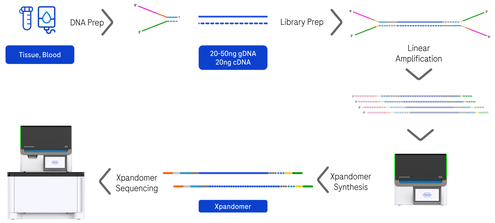



Fig 1. SBX-SL Workflow

This workflow is compatible with extracted DNA as well as already amplified cDNA libraries for RNA sequencing.

## Results

The extended read length offered by SBX-SL sequencing provides a range of advantages for genomic discovery, particularly in regions where traditional short-read sequencing is limited. SBX-SL can improve the detection of structural variants and enables more accurate long-range phasing of variants on the same haplotype. To demonstrate these capabilities, we benchmarked SBX-SL using well-characterized cell lines, including somatic tumor/normal pairs such as HCC1395 and H2009. As shown in Fig. 2, longer reads shows more evidence for expected SVs in DNA and RNA sequencing.

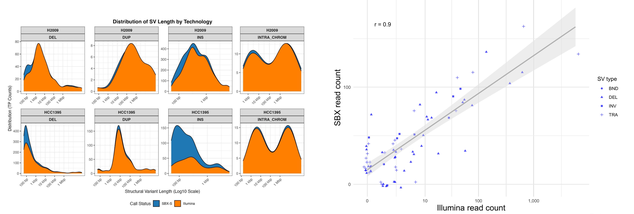






Figure 2. (left) break down of SV detection performance of SBX SL DNA on 2 well-characterized cancer cell lines.[3,4] (right) performance of SBX-SLR in measurements of SV expression from RNAseq (HCC1395).[4]

## Results

Variable Number Tandem Repeats (VNTRs) are short DNA sequences repeated a variable number of times in tandem at specific genomic locations. Their high variability between individuals makes them valuable for genetic studies, including disease association, forensics, and population genetics. However, due to their repetitive nature, short-read sequencing struggles to accurately genotype VNTRs in whole-genome sequencing (WGS) data. In contrast, the longer read lengths of SBX-SL sequencing can effectively span most VNTR regions, enabling accurate detection of VNTR expansions in the human genome.[5-7] Benchmarking on the HG002 reference cell line shows that SBX-SL provides better performance compared to SBX-D in detecting these events—particularly those larger than 100 base pairs (Fig. 3).

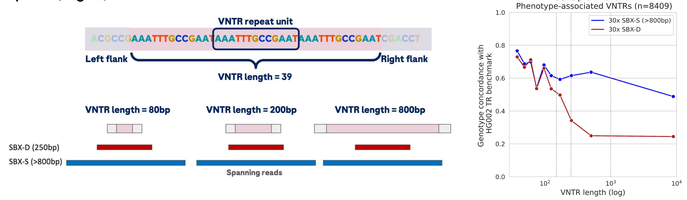






Figure 3. Accurate detection of VNTR variants requires long reads that can span VNTR sequence. SBX-SL can improve VNTR genotyping compared to SBX-D.

Variant phasing is the process of determining which genetic variants are inherited together on the same chromosome (haplotype). Long-read sequencing helps with phasing by spanning multiple variants within a single read, allowing accurate reconstruction of haplotypes over longer genomic distances. Benchmarking on HG002, SBX-SL is able to show better performance compared to short read sequencing specially in variants further apart than 500 bp (Fig. 4). Moreover, long range phasing can also improve detection of compound heterozygous variants.

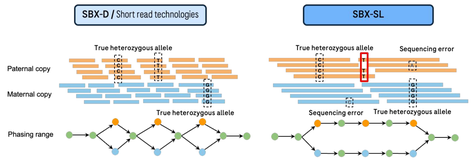
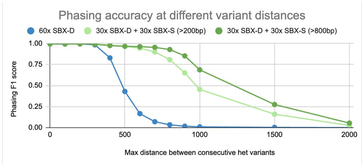
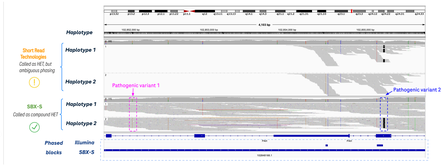



Figure 4. Schematic illustrating haplotype phasing using nearby heterozygous variants. The longer read lengths of SBX-SL enable robust phasing across greater genomic distances, resulting in longer contiguous haplotype blocks. Benchmarking on the HG002 reference genome shows that SBX-SL achieves higher phasing F1 scores, particularly for variant pairs separated by more than 500 bp. The bottom right panel highlights an example of a pathogenic compound heterozygous variant that was resolved only with SBX-SL, but not detected by short read sequencing.




## Results

Finally, SBX-SL is designed to enable multi-omics discovery by integrating longer sequencing from both DNA and RNA. As illustrated in Fig. 5, allele-specific RNA expression can be resolved through phasing of SBX-SL RNA reads. SBX-SLR accurately detected exonic variants seen in SBX-SLD; however, one allele is significantly over expressed.

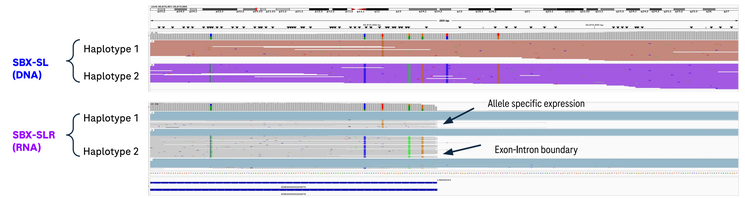



Figure 5. IGV snapshot illustrating multi-omics phasing using SBX-SL technology in the HG002 cell line. Reads were first phased using SBX-SL DNA data. RNA reads were phased and accurately assigned to paternal and maternal alleles. While SBX-SLR and SBX-SLD exonic variants show high concordance, SBX-SLR indicates strong allelic imbalance in expression of the second allele.

## Conclusion

In summary, SBX-SL showed improvement in the detection of large-scale germline and somatic events—including structural variants, tandem repeats, and long-range phasing—from both DNA and RNA sequencing. It serves as a powerful complement to SBX-D across whole-genome sequencing (WGS) applications. Combined, SBX-SL and SBX-D can leverage SBX-D accuracy and SBX-SL longer reads to enable broader genomics applications.

## Disclosures

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7. Method: adVNTR used for genotyping of pVNTR + gVNTR catalog in Javadzadeh, et al.

# Demonstrating the versatility, accuracy, and throughput of Sequencing By Expansion (SBX)

## Ultra fast whole genome sequencing from sample prep through variant analysis in less than 4 hours

**Jagadeeswaran Chandrasekar**,[1] Amal Chaturvedi,[2] Austin Doupnik,[2] Boone Hapke,[1] Brittany Kesic,[1] Chen Zhao,[2] Chuck Seberino,[2] Cynthia Cech,[1] Daniel Baker,[2] Dieter Heindl,[3] Dilmi Perera,[2] Doug Lopez,[2] Elise Le,[1] Emily Ormbrek,[1] Fong Chun Chan,[2] Hannes Kuchelmeister,[3] Jayalakshmi Rajaraman,[2] Joanne Leadbetter,[1] John Mannion,[2] Kendall Berg,[1] Mahdi Golkaram,[2] Maryam Rabiee,[2] Maryam Shenasa,[2] Matthew Lopez,[1] McKenna Osentowski,[1] Melanie Kirsche,[2] Melud Nabavi,[1] Nasim Farajpour,[1] Ryan Toma,[1] Salka Barrett,[1] Sam Bandara,[2] Taher Mun,[2] Taylor Lehmann,[1] TK Wasserman,[2] Won-Mean Lee,[2] Yui Umezawa,[1] Mark Kokoris[1]

[1]Roche Sequencing Solutions, Inc, Seattle, WA, USA; [2]Roche Sequencing Solutions, Inc, Santa Clara, CA, USA; [3]Roche Diagnostics GmbH, Penzberg, Germany

Poster no. 4092T

## Introduction to SBX-Fast

Sequencing By Expansion (SBX) technology is a novel sequencing approach that uses a biochemical process to encode the sequence of a target nucleic acid molecule into a measurable surrogate polymer called an Xpandomer.[1] SBX-Fast is duplex-based, amplification-free research workflow, designed with the intent to provide a deployable solution for rapid sequencing applications where time to result is important. This workflow produces high-accuracy results by linking both strands of the target DNA in a single sequencing read, enabling rapid and accurate identification of InDels, SNVs, STRs, and CNVs.

We evaluated SBX-Fast's variant-calling performance across 41 diverse cell lines from the Coriell Institute for Medical Research, which encompass a wide array of genetic conditions. Furthermore, in a speed trial to determine the minimum turnaround time, we successfully demonstrated the complete process—from library preparation to the final VCF variant file—in under four hours (3 hours, 59 minutes), using the HG002 reference sample.

## Materials and Methods

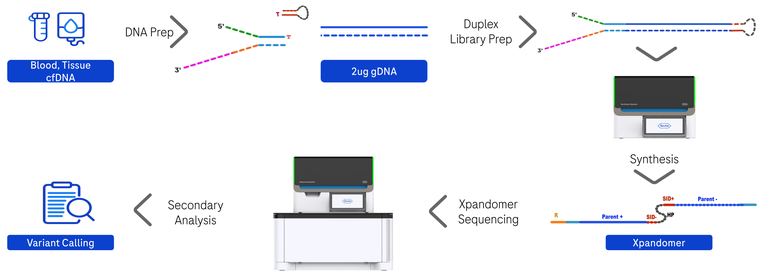
### Reference Benchmarking

2 µg of unsheared genomic DNA from each sample was input into the SBX-Fast workflow. Samples were sequenced as a trio for 1 hr.

### Time Trial

2 ng of unsheared genomic DNA from HG002 was input into a time optimized SBX-Fast workflow, and sequenced solo for 20 minutes to target >30x coverage.

**Figure 1. SBX-Fast Workflow**




SBX-D data were processed through SBX-optimized open source (XOOS) variant callers.[2-3]

## Accelerated Data Processing Overview

Raw sequencing data is processed (basecalling, demultiplexing, intramolecular consensus) in real-time on the sequencing system's integrated hardware. These consensus reads are then continuously mapped to the reference genome. Once the target coverage depth is achieved for a sample, a merged and sorted BAM file is generated. After the BAM files are generated, the Roche SBX Optimized Open Source (XOOS) tools are used for variant detection (SNVs, INDELs, CNVs, and STRs).

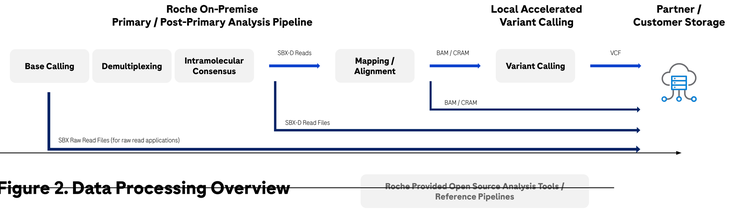



**Figure 2. Data Processing Overview**

## Reference Benchmarking (Results)

As shown in the table below, we successfully called the expected variant in all reference samples.

| Sample ID | Description | Variant Type | |
|---|---|---|---|
| NA25495 | Choroideremia | CNV | ✅ |
| NA23710 | Epileptic encephalopathy | CNV | ✅ |
| NA02325 | Translocated chromosome | CNV | ✅ |
| NA21698 | Chromosome deletion & CNV Reference | CNV | ✅ |
| NA08618 | Chromosome deletion | CNV | ✅ |
| NA03330 | Trisomy 13, Patau syndrome | CNV | ✅ |
| NA13480 | Williams–Beuren syndrome | CNV | ✅ |
| NA10283 | Hyperglycerolemia | CNV | ✅ |
| NA05117 | Duchenne muscular dystrophy | CNV | ✅ |
| NA22208 | Propionic acidemia | CNV | ✅ |
| NA04520 | Tuberous Sclerosis Complex 2 | CNV | ✅ |
| NA06151 | Machado-Joseph Disease | STR | ✅ |
| NA04079 | Friedreich's Ataxia | STR | ✅ |
| NA06894 | Fragile X mental retardation | STR | ✅ |
| NA03756 | Myotonic Dystrophy | STR | ✅ |
| NA13716 | Dentatorubral-Pallidoluysian Atrophy | STR | ✅ |
| NA23709 | Spinal and bulbar muscular atrophy | STR | ✅ |
| GM28741 | Homocystinuria-megaloblastic anemia | CNV | ✅ |
| GM27903 | Cerebral creatine deficiency syndrome 1 | SNV/INDEL | ✅ |
| GM28606 | Shwachman-Diamond syndrome | SNV/INDEL | ✅ |

| Sample ID | Description | Variant Type | |
|---|---|---|---|
| NA04327 | Duchenne muscular dystrophy | CNV | ✅ |
| NA23127 | Muscular Dystrophy, Becker Type | CNV | ✅ |
| NA06804 | Lesch-Nyhan syndrome | CNV | ✅ |
| NA09834 | Basal Cell Nevus Syndrome | CNV | ✅ |
| NA12214 | Charcot-Marie-Tooth disease type 1A | CNV | ✅ |
| NA05876 | DiGeorge syndrome | CNV | ✅ |
| NA22010 | Propionic Acidemia, clinically affected | SNV | ✅ |
| NA22011 | Clinically unaffected mother | SNV | ✅ |
| NA22012 | Clinically unaffected father | SNV | ✅ |
| NA14553 | Arterial Calcification | SNV | ✅ |
| NA00882 | Fabry Disease | SNV | ✅ |
| NA00372 | Gaucher Disease, Type I | SNV/InDel | ✅ |
| NA22113 | Propionic Acidemia clinically affected sister 1 | Delins | ✅ |
| NA22112 | Propionic Acidemia clinically affected sister 2 | Delins | ✅ |
| NA22111 | Clinically unaffected mother | Delins | ✅ |
| NA22110 | Clinically unaffected father | Delins | ✅ |
| NA11195 | Phenylketonuria | SNV/InDel | ✅ |
| NA23391 | Myotonic Dystrophy | STR | ✅ |
| NA05131 | Fragile X | STR | ✅ |
| NA16202 | Clinically unaffected mother | STR | ✅ |
| NA16203 | Friedreich's Ataxia | STR | ✅ |

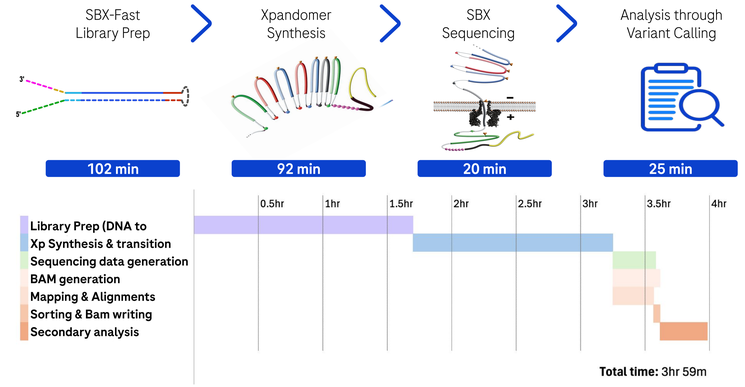
**Table 1. Reference Samples Results**

## Time Trial (Results)

Using HG002, we demonstrated DNA to VCF in 3 hours, 59 minutes.

Library prep through VCF in **3 hours, 59 minutes**

| Sample | Total Duplex Reads (B) | Mean Insert Read Length | Coverage | % genome at >10x coverage |
|---|---|---|---|---|
| HG002 | 1.06 | 158 | 33x | 99.5 |







**Total time:** 3hr 59m

## Conclusion

For settings requiring rapid genomic analysis, SBX-Fast has the potential to deliver timely information that could impact decisions to improve outcomes.

## Disclosures

This study was funded by Roche Diagnostics International Ltd (Rotkreuz, Switzerland)

All authors are employees of Roche Sequencing Solutions, Inc. or Roche Diagnostics GMBH and may hold non-voting equity securities in F. Hoffmann-La Roche Ltd.

## References

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# Whole Genome Sequencing Minimum Residual Disease Detection using Sequencing By Expansion (SBX)

**Kendall Berg,**[1] Thao Ho,[1] Abid Hasan,[2] Alan Kimura,[1] Alberto Gatto,[2] Alec Sautter,[1] Alex Lehmann,[1] Anasha Arryman,[1] Chen Zhao,[2] Cynthia Cech,[1] Dinendra Abeyawardhane,[1] Emily Ormbrek,[1] Fan Song,[2] Grant Kingsley,[1] Jagadeeswaran Chandrasekar,[1] Joanne Leadbetter,[1] John Mannion,[2] Lacey McGee,[1] Mahdi Golkaram,[2] Marc Prindle,[1] Matthew Lopez,[1] McKenna Osentowski,[1] Megan Freer,[1] Megan LeProwse,[1] Melud Nabavi,[1] Robert Busam,[1] Ron Cicero,[2] Ryan Toma,[1] Taylor Lehmann,[1] Thomas Reid,[1] Upneet Bala,[1] Mark Kokoris[1]

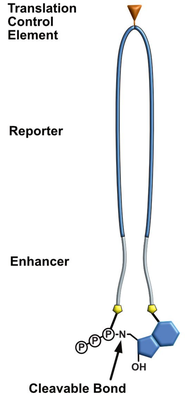
[1]Roche Sequencing Solutions, Inc, Seattle, WA, USA; [2]Roche Sequencing Solutions, Inc, Santa Clara, CA, USA

Poster no. 4035T

## Background

Detection of minimal residual disease (MRD) is one of the strongest predictors of cancer relapse post treatment, and therefore a critical component of cancer management. While WGS of cfDNA offers a non-invasive and comprehensive view, it is limited by the error rate and sensitivity required to detect mutations especially in low tumor burden conditions.[1] Here, we assess the performance of WGS workflow for MRD detection using Sequencing by Expansion (SBX).

## Introduction to SBX




Sequencing By Expansion (SBX) technology is a novel sequencing approach that uses a biochemical process to encode the sequence of a target nucleic acid molecule into a measurable surrogate polymer called an Xpandomer.[2]

**Figure 1. X-NTP Structure**

The Expandable Nucleotide Triphosphate (X-NTP) is the building block of the Xpandomer. There are 4 different XNTPs, one for each nucleotide. Highly designed components of the XNTP allow for high signal to noise and non-stochastic translocation control.

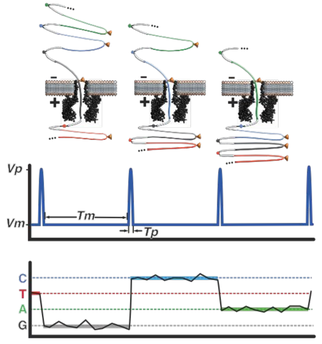



**Figure 2. Xpandomer Measurement**

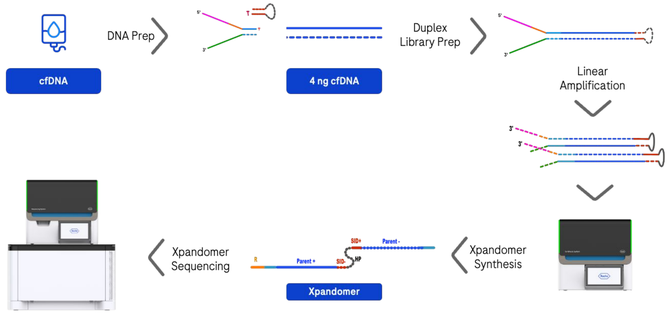
The SBX workflow has two components: synthesis and measurement. During synthesis, X-NTPs are used to enzymatically transcribe the template DNA into an Xpandomer molecule.

After synthesis, the resulting Xpandomer is sequenced by stepwise translocation through a nanopore.

## SBX-D Workflow

SBX-D is a library prep and linear amplification sequencing workflow that achieves high accuracy by linking both strands of the target DNA within a single sequencing read. This allows for single-strand errors that arise on separate strands of the DNA molecule to be resolved, thereby achieving higher accuracy.

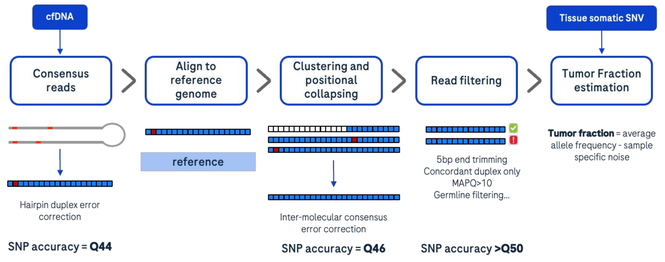
**Fig 3. SBX-D workflow for MRD**




## Materials & Methods

Fifteen (15) healthy and fifteen (15) cancerous cfDNA research samples from a diverse cohort of cancer diagnoses and stages were used to benchmark MRD detection performance. Four nanograms (4 ng) of DNA was input into the SBX-D library prep workflow and sequenced targeting 100x coverage. SBX data were processed through SBX-optimized open source (XOOS) variant callers.[3]

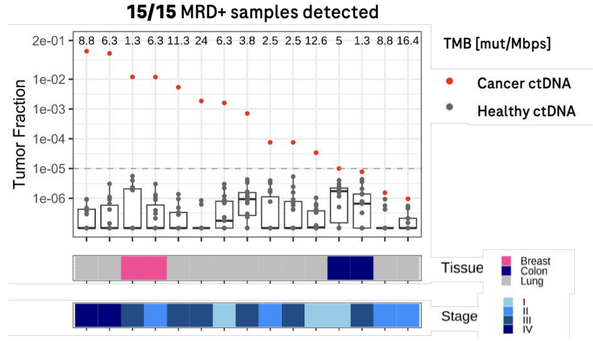
**Fig 4. Read Processing and Analysis**




## Results

MRD signal was detected for all cancer sample driven cfDNA. Using inter-molecular consensus formation on duplex reads and bioinformatic error correction, we observed SNP accuracy >Q50, and detected low frequency variants indicative of MRD in all cancer samples, even in a sample with tumor fraction as low as 1x10-6. Average measured tumor fraction in healthy cfDNA samples was measured to be 6x10-7.

**Fig 5. MRD detection**




## Conclusion

This preliminary result demonstrates the potential of SBX-D for minimal residual disease (MRD) applications, even with challenging samples. This enhanced sensitivity could enable earlier and more precise identification, which may lead to more timely and tailored treatment to improve long-term outcomes.[1]

## Disclosures

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## References

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2. https://doi.org/10.1101/2025.02.19.639056
3. https://roche-axelios.gitbook.io/xoos

# Whole Genome Precision at Scale: Sequencing by Expansion for Cancer Genomics Research

**Mahdi Golkaram,[2]** Alan Kimura,[1] Alec Sautter,[1] Alex Lehmann,[1] Amirhossein Afshinfard,[2] Anasha Arryman,[1] Brent Banasik,[1] Brittany Kesic,[1] Chen Zhao,[2] Cynthia Cech,[1] Dinendra Abeyawardhane,[1] Erfan Sayyari,[2] Fan Song,[2] Fong Chun Chan,[2] Geoffrey Barrall,[2] Grant Kingsley,[1] Hasham Saqib,[2] Jagadeeswaran Chandrasekar,[1] John Mannion,[2] Ke Bi,[2] Ke Tang,[2] Kendall Berg,[1] Kevin Smith,[1] Majid Babazadeh,[1] Megan Freer,[1] Melanie Kirsche,[2] Mike Molnar,[2] Roseann Ahlin,[2] Taher Mun,[2] Thomas Reid,[1] Mark Kokoris[1]

[1]Roche Sequencing Solutions, Inc, Seattle, WA, USA; [2]Roche Sequencing Solutions, Inc, Santa Clara, CA, USA

Poster no. 4073T

## Background

Whole genome sequencing (WGS) offers a more comprehensive view of the cancer genome compared to targeted methods, enabling the detection of several genetic alterations in a single test. However, the substantial sequencing requirements, challenges in achieving high sensitivity while maintaining low false positive rates—particularly in low-quality samples like Formalin-Fixed Paraffin-Embedded (FFPE) tissues—and long turnaround times have hindered broader adoption of WGS.[1]

Sequencing By Expansion (SBX) offers flexible, ultra-high-throughput sequencing with rapid turnaround.[2] Here, we show that hairpin duplex SBX (SBX-D) can overcome these barriers, enabling broader WGS adoption in oncology research. We present advanced bioinformatics algorithms for accurate detection of somatic variants, including SNVs, INDELs, copy number alterations (gain, loss, LOH), structural variants, and biomarkers of interest such as tumor mutational burden, mutational signatures, HLA typing.

## Materials and Methods

In this study, we performed 70–100x for both SBX (mean) and Illumina WGS (mean) on 20 FFPE tissues using 100 ng DNA across a range of sample qualities to benchmark SBX variant calling against Illumina NGS.

Illumina samples were sequenced via NovaSeq 6000 S4 and processed through DRAGEN 4.3 using Illumina recommended parameters.[3] SBX-D data were processed through SBX-optimized open source (XOOS) variant callers.[4-5]

Overall, SBX-D approach shows higher SNP and INDEL accuracy compared to Illumina. Better SNV and INDEL accuracy could be attributed to duplex sequencing and replacing PCR with linear amplification which can significantly reduce long HP error rate (Fig. 1).

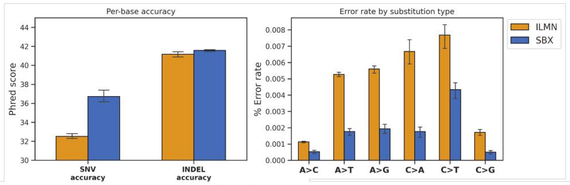


Figure 1. Comparison of SNV and INDEL accuracy between Illumina and SBX-D sequencing of FFPE tissues. Duplex sequencing and linear amplification contribute to higher SNV and INDEL accuracy in SBX-D.

## Disclosures

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## Results - Variant Calling

Overall, we observed high alignment between Illumina and SBX-based variant calling (Fig. 2). Duplex sequencing and the replacement of PCR with linear amplification in SBX reduced SNV and INDEL error rates, improving sensitivity for detection of subclonal variants and variants in challenging genomic context, such as low-complexity regions. These gains were achieved while maintaining the same low false positive rates as Illumina (0.13 SNV FP/Mbp and 0.08 INDEL FP/Mbp).

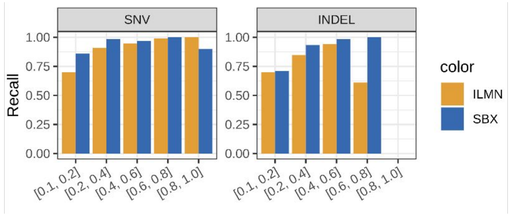


Figure 2. Performance of SNV and INDEL calling in SBX-D sequencing of FFPE tissues. Duplex sequencing and linear amplification contribute to high SNV and INDEL accuracy in SBX-D

In addition to small variant calling, next we evaluated the performance of SBX-D based sequencing to perform somatic allele specific copy number variant calling, somatic structural variant calling and HLA typing. Overall, we observed >99% concordance in our cohort consisting of 20 T/N pairs (Fig. 3).

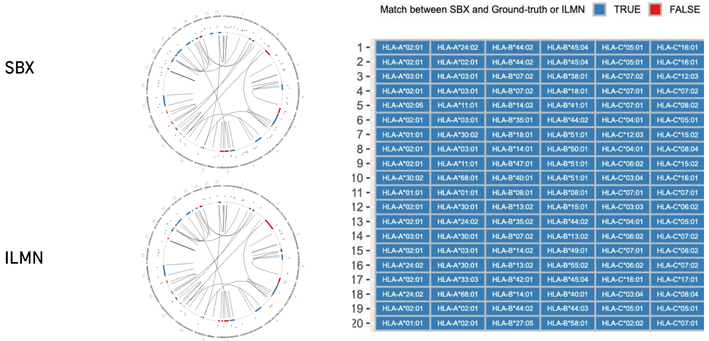


Figure 3. Benchmarking of comprehensive genomic profiling using SBX-D including small variant calling (SNV/INDEL), allele specific copy number variants (CNV), structural variants (SV) and HLA typing. Circos plots display COSMIC small variants using purple dots, CNVs on inner tracks displayed with red (gain), blue (loss), orange (LOH), and curved arcs to represent larger SVs such as translocations, inversions, insertions, deletions, or duplications across genomic regions.

## Results - Biomarkers

Finally, we evaluated the performance of SBX-D in detection of biomarkers. Overall, SBX-D shows a strong performance across all measured biomarkers suggesting SBX-D as a reliable approach for biomarker research studies (Fig. 4).

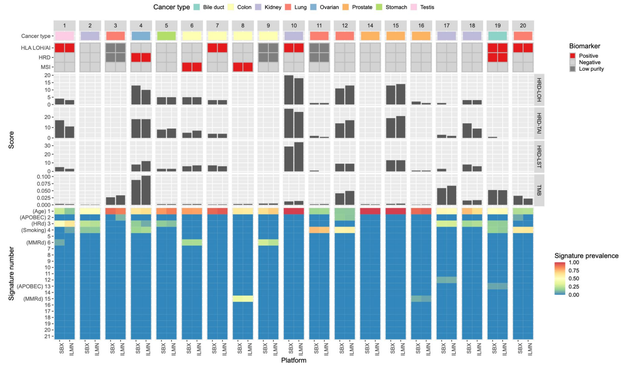


Figure 4. Comprehensive benchmarking of SBX-D base oncology biomarkers from WGS of FFPE samples. Results show a high concordance across a variety of biomarkers including tumor mutational burden (TMB), microsatellite instability (MSI), homologous recombination deficiency (HRD), HLA loss or allelic imbalance (LOH/AI), and mutational signatures.

## Conclusion

Here, we benchmarked comprehensive genomic profiling and biomarker measurements using WGS of FFPE tissues using SBX-D technology. Overall, SBX-D showed highly consistent results across all benchmarking samples and variant types, while also detecting low AF subclonal events due to a high base accuracy.

SBX-D demonstrates reasonable sensitivity and turnaround times, offering a promising approach for accurate whole genome sequencing in oncology research applications.

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