



OncoDNA[®]
THE CANCER THERANOSTIC COMPANY

International User Group Meeting

22nd and 23rd of April 2026

Insights from OncoDEEP validation on the AVITI24 platform

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About PathoNext

- Founded in Leipzig (Germany) in 2015 by shareholders with a longtime expertise in the field of molecular diagnostics
- Highly experienced team specialized in NGS
- Trusted partner for **molecular pathology**, working with hospitals and institutes of pathology, including **molecular tumor boards**
- Cooperations with other labs and various universities in the field of **human genetics** (especially cardiogenetics) and **metagenomics**



Our Expertise

Wet Bench

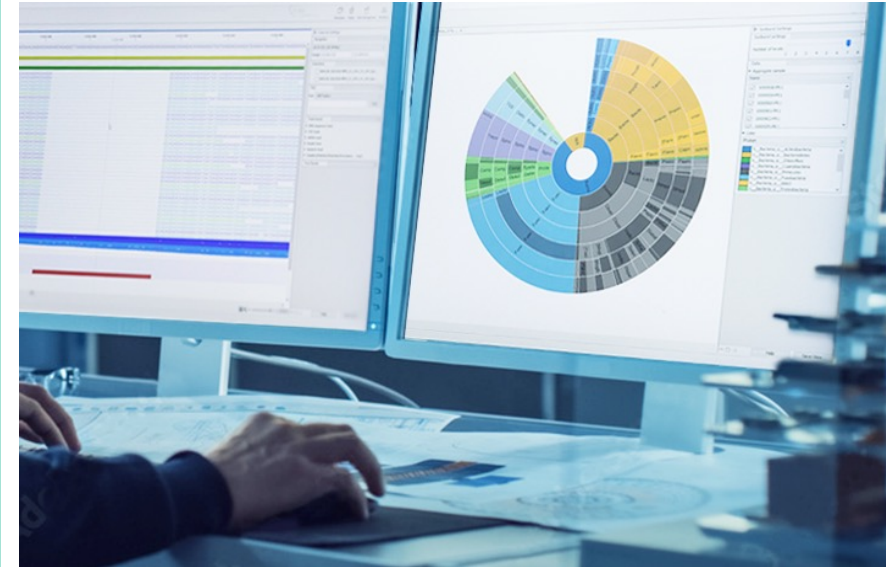
- Automated DNA and RNA extraction from a variety of starting materials
 - Especially from difficult samples (FFPE slides, paper tips)
- Automated NGS library preparation for WGS, WES, and panels
- Sequencing instruments:
 - Illumina NovaSeq 6000, NextSeq 500, MiSeq
 - Element Biosciences AVITI24 (since October 2025)
- Certified routine laboratory (**DIN ISO 17025**)
- Trusted NGS **ProLab for Twist Biosciences**
- Successful participation in beta testing projects with global companies like Illumina and Roche



Our Expertise

Data analysis and Interpretation

- Secondary data analysis for WGS/WES/panel and metagenomics sequencing data
- Interpretation of therapeutically relevant variants for clinical reporting according to:
 - **ACMG** guidelines (germline) and
 - **ClinGen** and Cancer Genomics Consortium (somatic)
- Development of customized NGS-Panels
- Big Data analysis with various software platforms:
 - Qiagen CLC Genomics Server/Workbench
 - Illumina's DRAGEN
 - varvis



Quality Management

- Extensive experience in quality assurance

The successful accreditation according to **DIN ISO 17025** confirms our high-quality standards .

- The regular and successful participation in international round robin tests for somatic mutations (EMQN, European Molecular Genetics Quality Network) to prove the implementation of quality management.



EMQN  **RfB**
Referenzinstitut für Bioanalytik
Delivered in collaboration with our partner
Reference Institute for Bioanalytics (RfB) in
compliance with the RfBÄK part B5

Certificate of Participation 2023

This is to certify that
PathoNext GmbH

Participated in 2023 in the following EMQN CIC external quality assessment schemes:

Scheme	Genotyping	Interpretation	Clerical	Result
ONCOGENE PANEL	2.00			Satisfactory

The laboratory participated in 1 scheme and passed 1 of them. The detailed performance data are given on the Individual Laboratory Report (ILR). When viewing the certificates, you should ensure that 1 scheme is listed.

Key: > Scheme Mean < Scheme Mean Poor performance
NRS: No Results Submitted **WFS:** Withdrew From Scheme

Signed by: 
Prof. David Barton
Chair of SSAB


Dr. Simon Patton
Managing Director

EMQN CIC is a community interest company which provides external quality assessment schemes for genomic diagnostic testing laboratories.
EMQN is a UKAS accredited proficiency testing provider, No. 4367 and is based in Manchester, UK.

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New Developments in our Lab

Motivation:

- Cost-efficiency to remain competitive
- Expand our CGP capabilities
- Reduce the dependency towards a single provider



Looking for alternatives...

Element Biosciences - AVITI24

- Multiomics platform, delivering high quality and affordable sequencing.
- Short read sequencing, flexible throughput
- Avidite Base Chemistry
- Rolling circle amplification (PCR-free)
- Cloudbreak Freestyle for loading linear libraries from existing library prep solutions



Advantages of AVITI24

Observations from sequencing custom panel libraries:

Parameter	NovaSeq	Aviti24
Q30	90.6	92.7-94.95
Mapping rate	96-98%	>99%
Mapped single reads	3-7%	1.5-2.85%
Duplicates	~50%	28-35%
Coverage		1.5x higher coverage with comparable reads
Variant Calls		All relevant variants detected with comparable AF (>4%)

We made these observations also in other types of libraries (WES, LBx, shotgun metagenomics etc.)

Advantages of AVIT124

- **Improved Q30, slightly better mapping rates**
- **Impressive reduction of duplicate levels**
- **Reaching the same coverage depth with way less data**
- **Cost-efficient and flexible**

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Challenges:

- Reaching the expected output considering very heterogeneous runs (e.g. 5 different library types)
- More susceptible to adapter content in the libraries (like for FFPE derived libraries)
- Adapting routine processes to longer sequencing times (29-31h instead of 24h)

AVITI24 compatible solution for CGP

- 20% of our NGS samples require comprehensive genomic profiling.
- Our previous solution (Illumina TruSight Oncology 500) required additional steps to enable the sequencing on AVITI24.

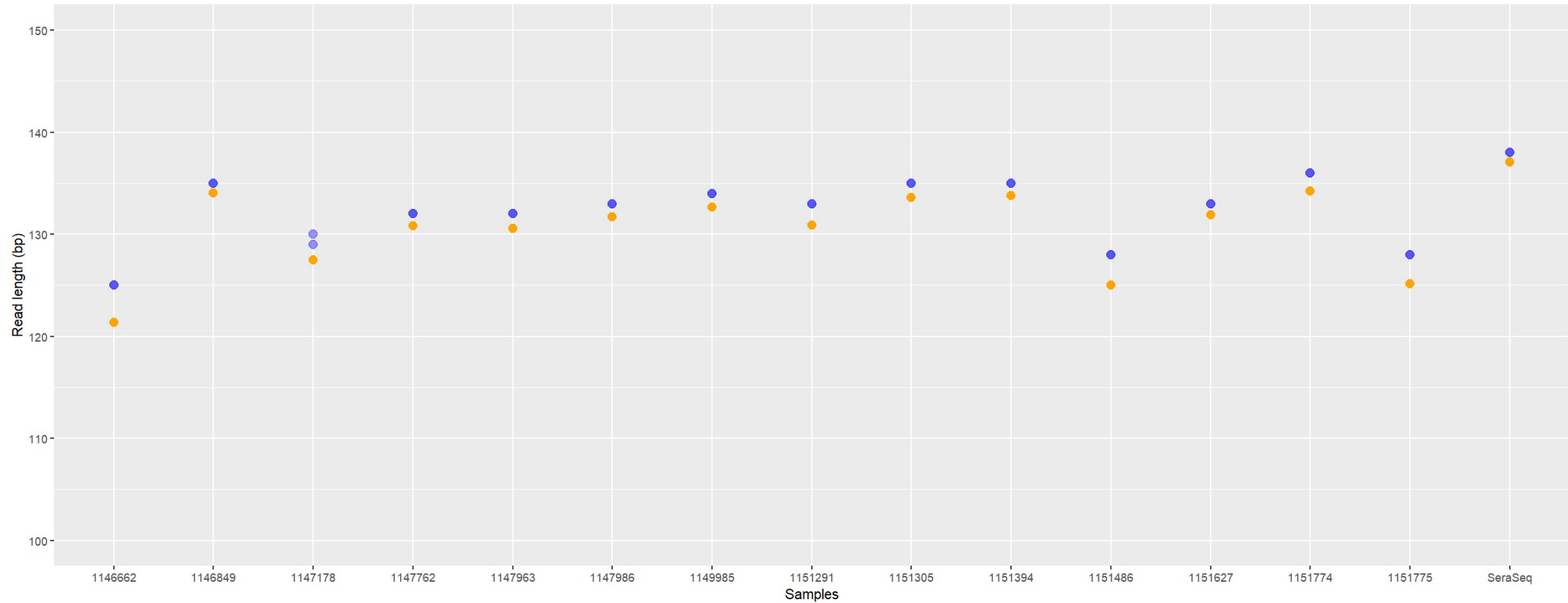
OncoDEEP

- Comprehensive panel of 638 genes, allowing for SNV and indel detection, CNVs, and fusions at DNA level
- Genomic signatures LOH, MSI, TMB (HRD soon)
- RNA fusion panel (especially the large RNA fusion panel is of high interest)
- BioIT included (OncoKDM), reports validated by OncoDNA
- Familiar Twist chemistry

Validation plan

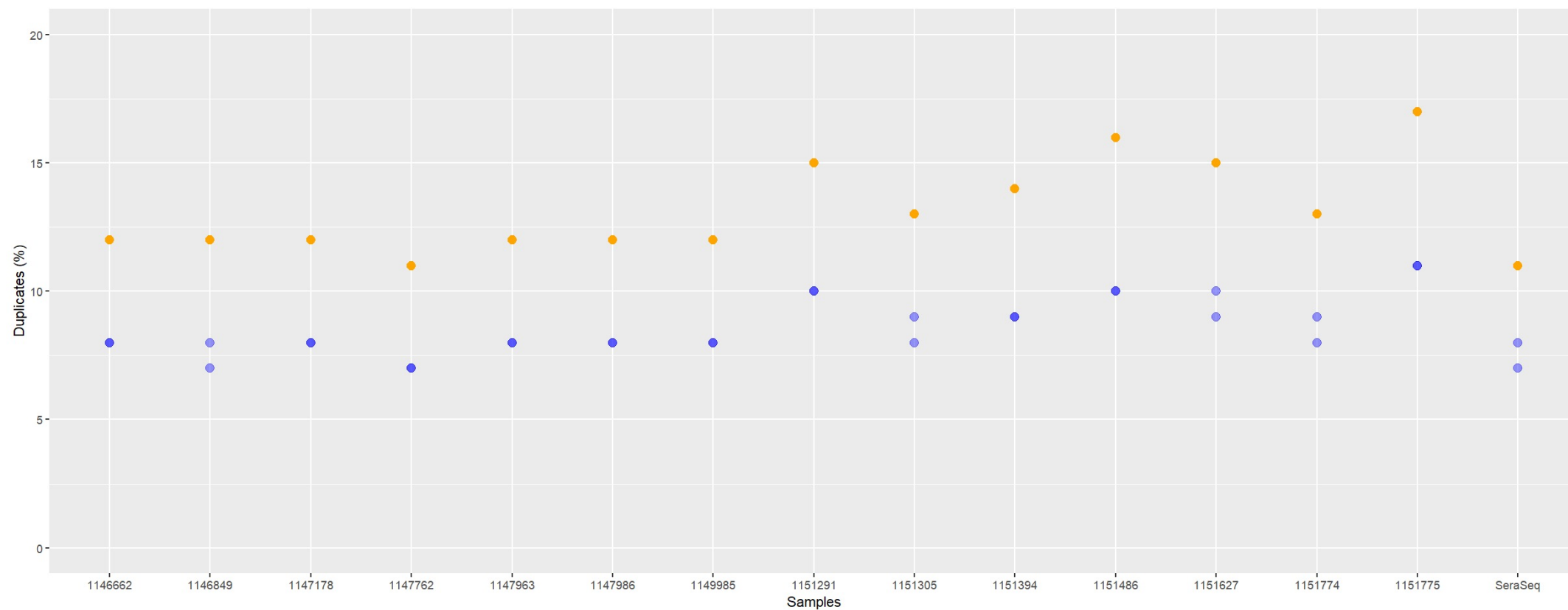
- **Samples:**
 - 16 DNA samples (15 prior characterized samples, one reference sample – SeraSeq DNA Mix v3) from various entities and of various quality (**DIN: 2.1- 6.2**)
 - 16 RNA samples (prior characterized)
- **Library preparations** on two different days
- **Sequencing** on two independent AVITI24 runs:
 - to assess the reproducibility
 - lanewise on one of the runs, to assess within-run concordance
- **Analysis with OncoKDM**

Results –Read length



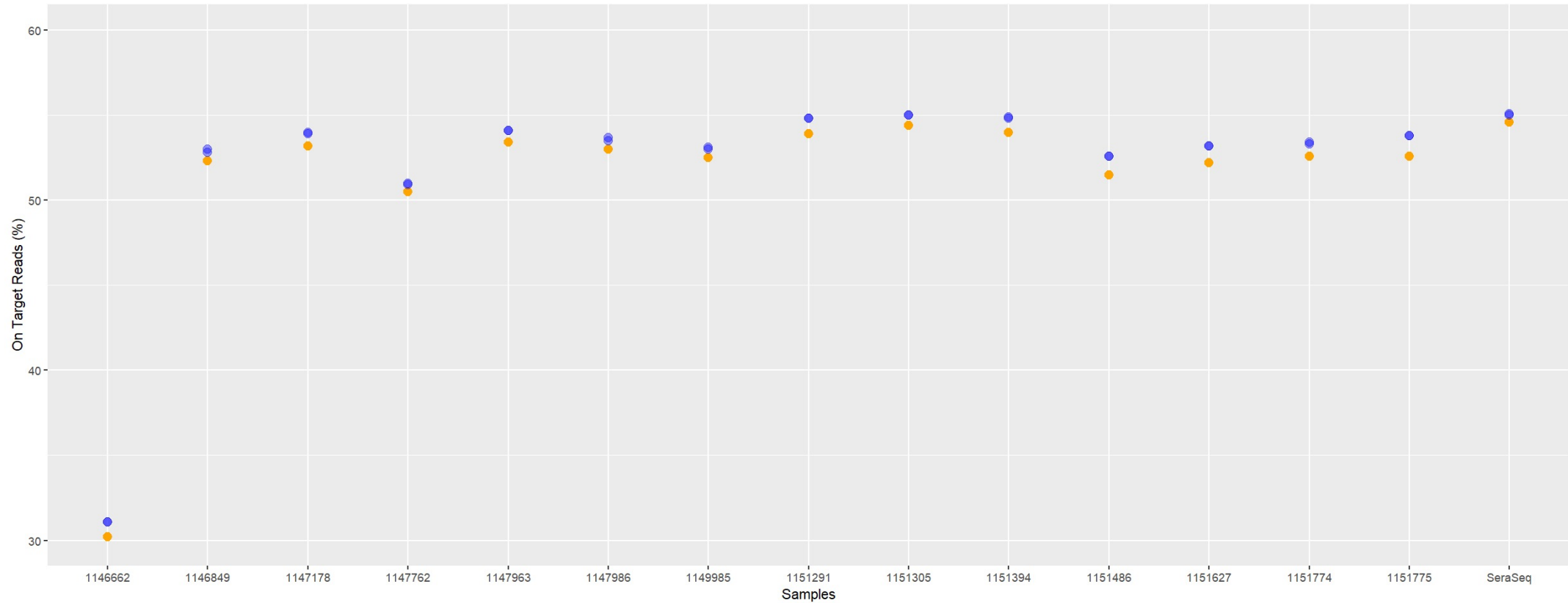
- The length of the reads varies because of adapter clipping and correlates with the quality of the sample.

Duplication Rates



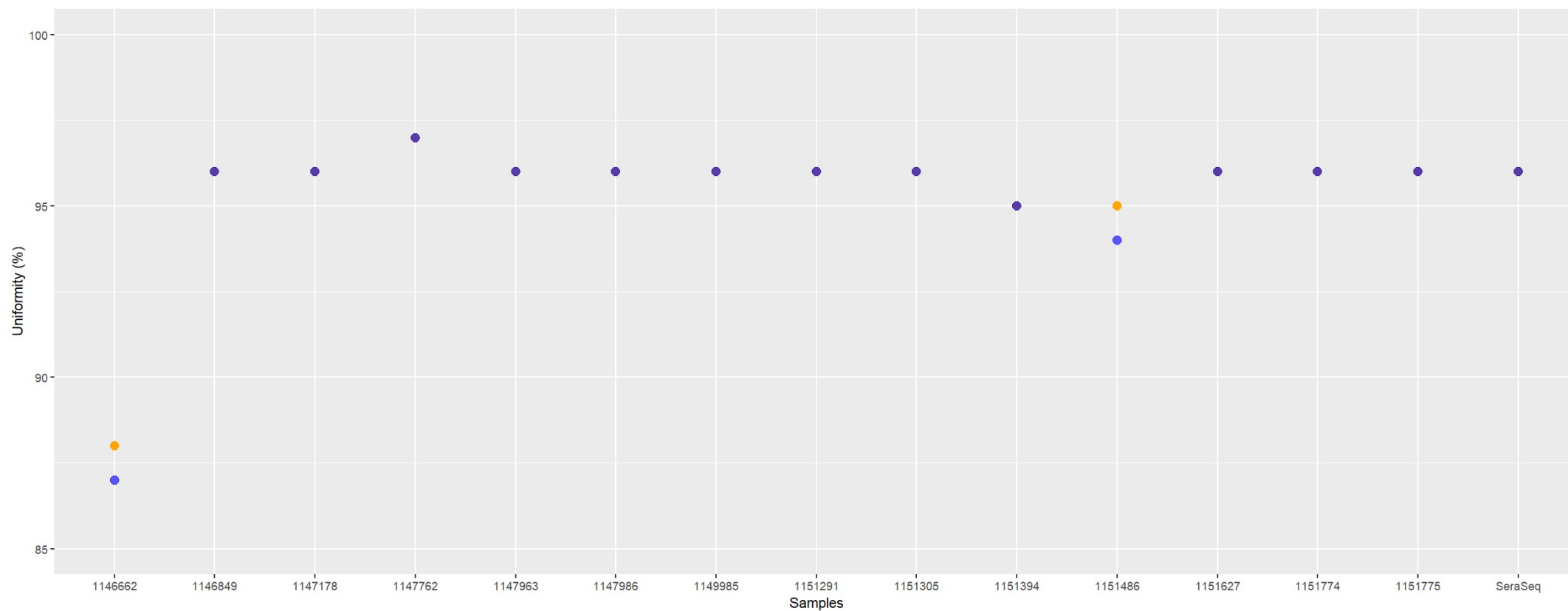
- Low duplication levels compared to our prior experience

On Target Rates



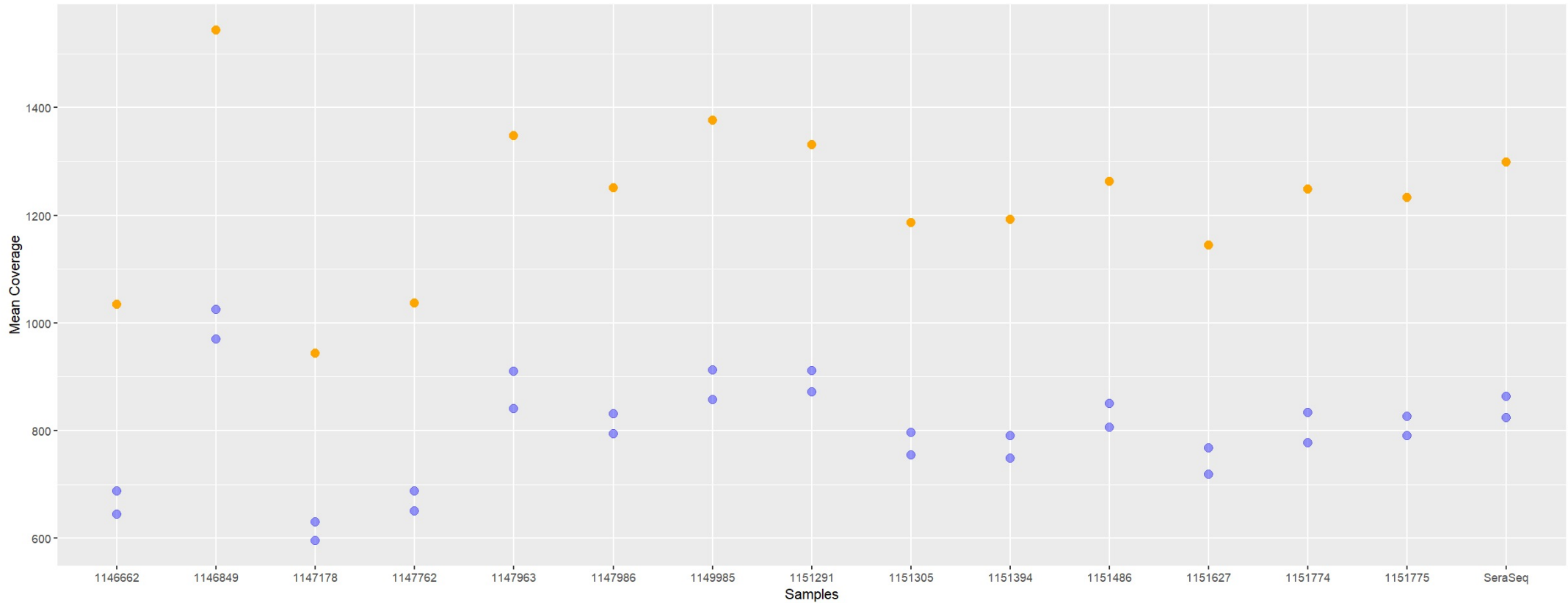
- Constant on target rates across samples and runs
- Outlier is a very degraded sample.

Uniformity



- Same uniformity between runs
- Outlier is a very degraded sample.

Mean Coverage



- Extremely good coverage, even with lower data amount.
- The data amount per sample in the „orange“ run is ~twice as high as in the „blue“ run.

Performance

- **Sensitivity of 98% for SNVs** based on the reference sample and the prior characterized samples (TSO500)
- Very good agreement of the **TMB and MSI** values with the expectations
- **11 out of the 12 expected CNVs** in the reference sample have been detected.
- Discrepancies were especially in variants with $AF < 4\%$
- Specific expected fusion events could not be detected because they are not covered yet by the RNA panel, but should be detectable with the large panel.

Ongoing work

- We are at the beginning of our OncoDEEP journey!
- Adapt our process to the new TAT of 10-15 business days, but we see great potential to reduce it.
- Optimizing the required sequencing output per sample
- Comparison of a larger cohort of samples characterized prior with TSO500

Key Take Away Slide



1. OncoDEEP performs robustly on AVITI24 and the synergy between sequencing platform and assay has reduced our hands-on time in the wetlab
2. We are reaching higher coverages and a better uniformity of the coverage with less sequencing data per sample.
3. OncoDEEP on AVITI24 is a flexible and cost-efficient solution for CGP (We were able to reduce our costs by ~ 35%).
4. Longer sequencing runtime and dry lab part lead currently to longer turn-around times, but we look forward to finetuning our workflow, especially having the constant support from OncoDNA.

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