



## European User Group Meeting OncoDEEP® Kit

21<sup>st</sup> - 22<sup>nd</sup> October 2024

### Development, design and validation of the OncoDEEP Kit

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## Plan

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- OncoKDO: Our hidden treasure
- Future Directions of the OncoDEEP Kit
- OncoDEEP Kit:  
The SEGLH Optimisation and Validation Experience

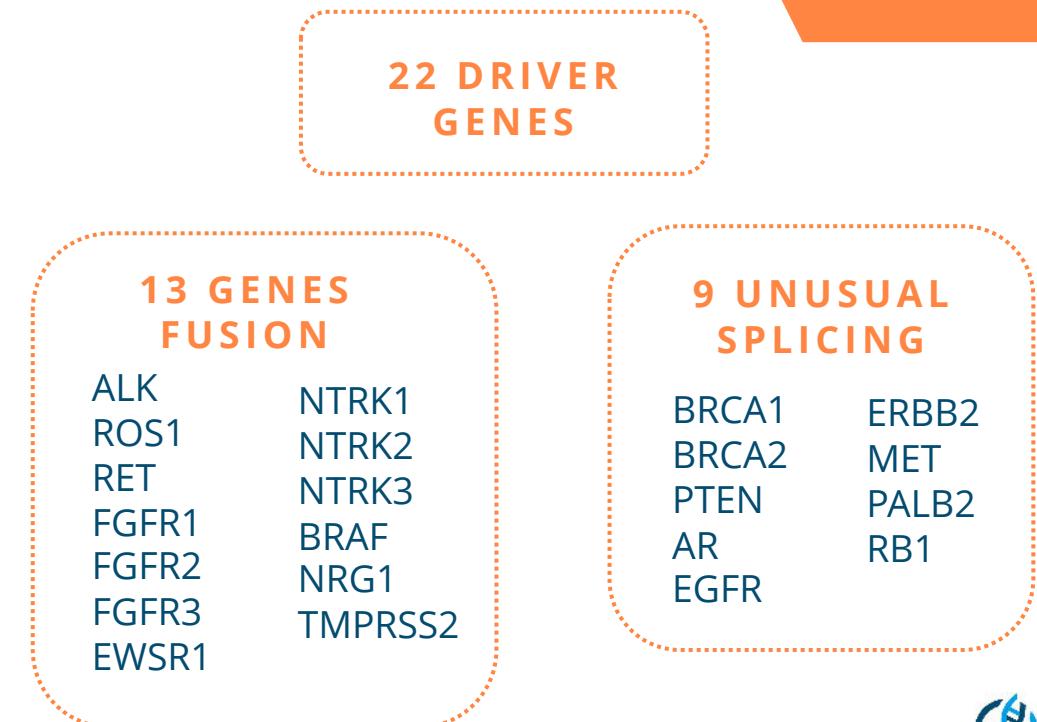
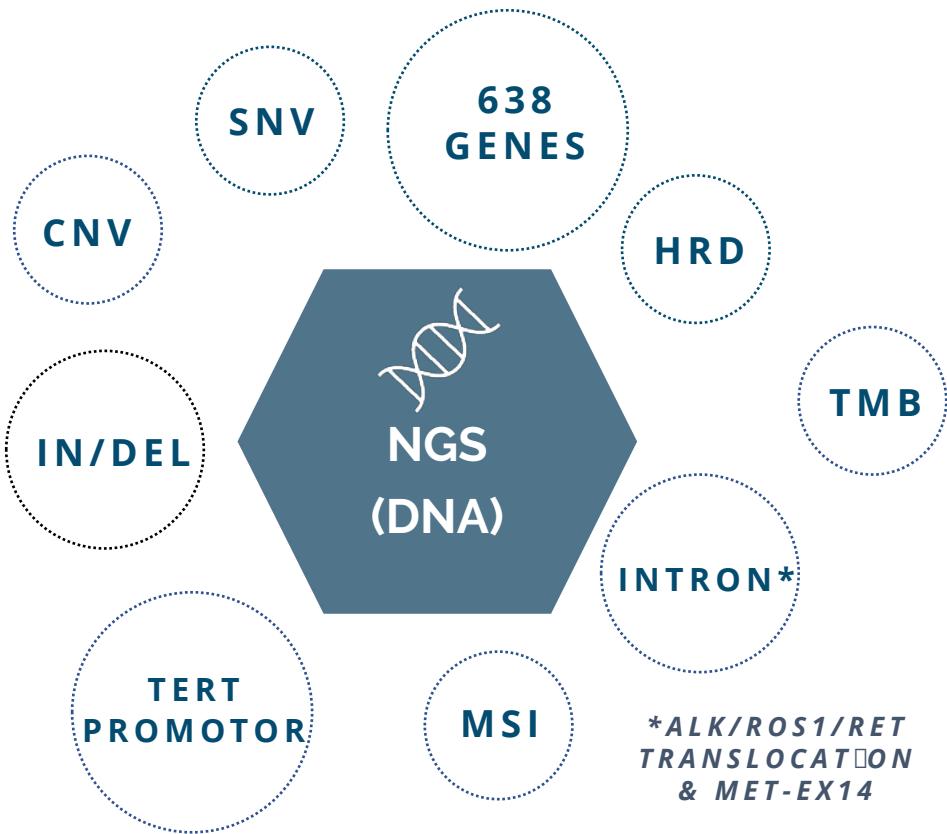
# Introduction

Comprehensive genomic profiling (CGP) is increasingly used in routine clinical practice, especially in oncology. Here are some key uses of CGP:

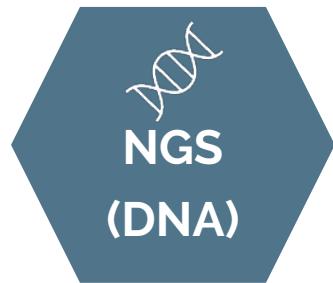
- 1. Personalized Treatment:** CGP helps identify specific mutations and alterations in a patient's tumor, guiding the selection of targeted therapies and immunotherapies that are more likely to be effective.
- 2. Diagnosis and Prognosis:** It can aid in the diagnosis of rare cancers or subtypes by revealing genetic changes that define the disease. Additionally, certain genomic alterations can provide prognostic information, helping predict disease progression.
- 3. Identifying Resistance Mechanisms:** CGP can uncover mutations that confer resistance to specific therapies, enabling oncologists to adapt treatment strategies proactively.
- 4. Clinical Trials:** Patients with specific genetic alterations identified through CGP may qualify for clinical trials targeting those mutations, providing access to novel therapies.
- 5. Hereditary Cancer Risk Assessment:** Beyond tumor profiling, CGP can identify germline mutations that may indicate a hereditary predisposition to cancer, informing patients and their families about risk management strategies.

# Design and Content Considerations

The **OncoDEEP** panel contains the most relevant and complete cancer gene panel. Over the time this panel was optimized to include **all clinically-relevant oncology targets**. The panel is composed of **638 genes**, reporting genomic alterations (SNV, insertion, deletion, CNV) and complex genetic signature (**HRD, MSI and TMB**).

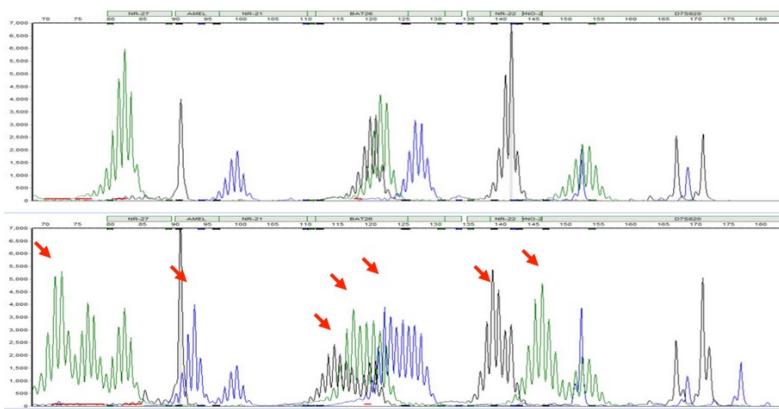


# Design and Content Considerations



- Approx 150 genes directly linked to treatment option accros different cancer types
- Content linked to relevance to cancer biology, emerging biomarkers and biomarkers linked to clincial trials : deep analysis of scientific publications
- Includes Oncogenes and Tumor suppressor genes, genes involved in cell growth, division, apoptosis, and DNA repair.
- Genomic signatures:

## 1. MSI (Microsatellite Instability)



1164 Loci distributed among the genome

NGS-based methods demonstrate superior performance to previous technologies

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC7010320/>

State: **Stable**

Score -0.3008333237648852

State Stable

PercentInstability 4.59%

Positif\_Markers 38/828

FAIL\_Markers 336/1164

MARKER LABEL STATUS

BAT-25 STABLE

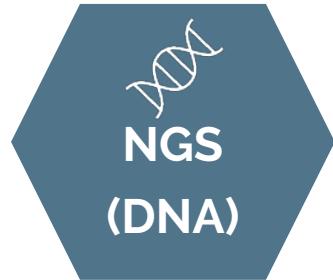
BAT-26 STABLE

NR-21 STABLE

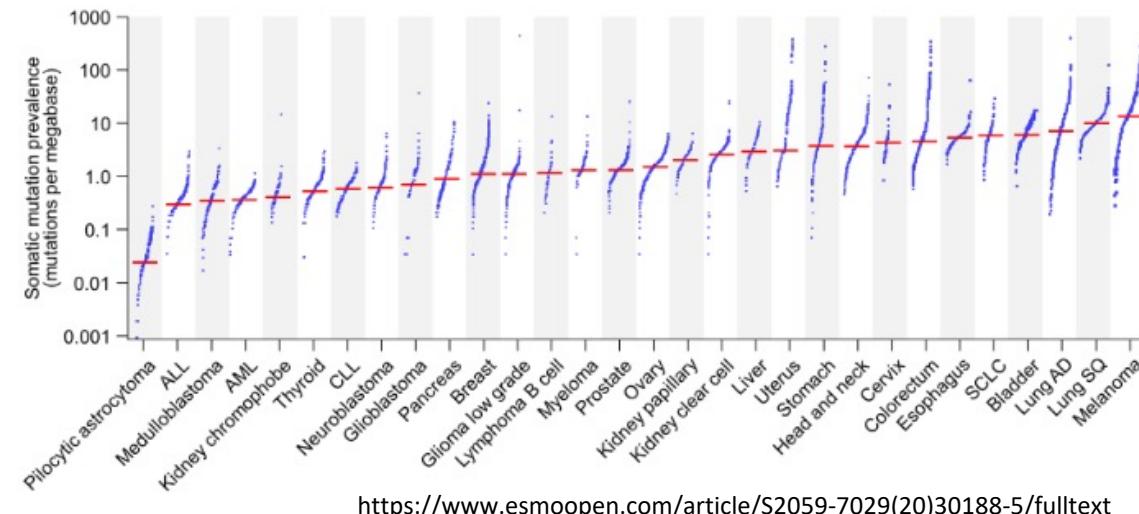
NR-27 STABLE

D2S123 STABLE

# Design and Content Considerations



- Genomic signatures:
- 2. TMB (Tumour Mutational Burden)



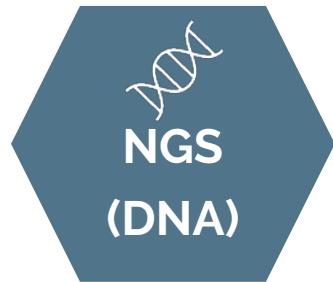
[https://www.esmoopen.com/article/S2059-7029\(20\)30188-5/fulltext](https://www.esmoopen.com/article/S2059-7029(20)30188-5/fulltext)

At a minimum, sequencing around **1-2 Mb** is considered necessary to obtain a TMB estimate  
(OncoDEEP KIT is 1.7/1.8 Mb)

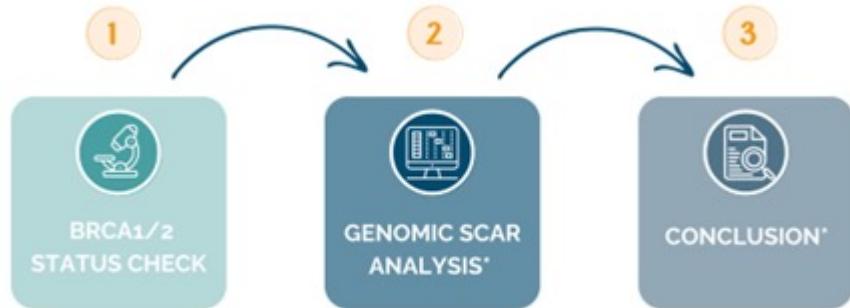
Number of mutations is assessed considering only the SNVs that are VUS (>10%) and excluding all pathogenic/likely pathogenic variants, specific germline mutations (based on gnomad), synonymous mutations, polymorphisms (based on gnomad), low coverage (80X) are excluded.

% of tumoral cells crucial to have a good estimation of TMB

# Design and Content Considerations



- Genomic signatures
- 3. HRD (Homologous Recombination Deficiency)



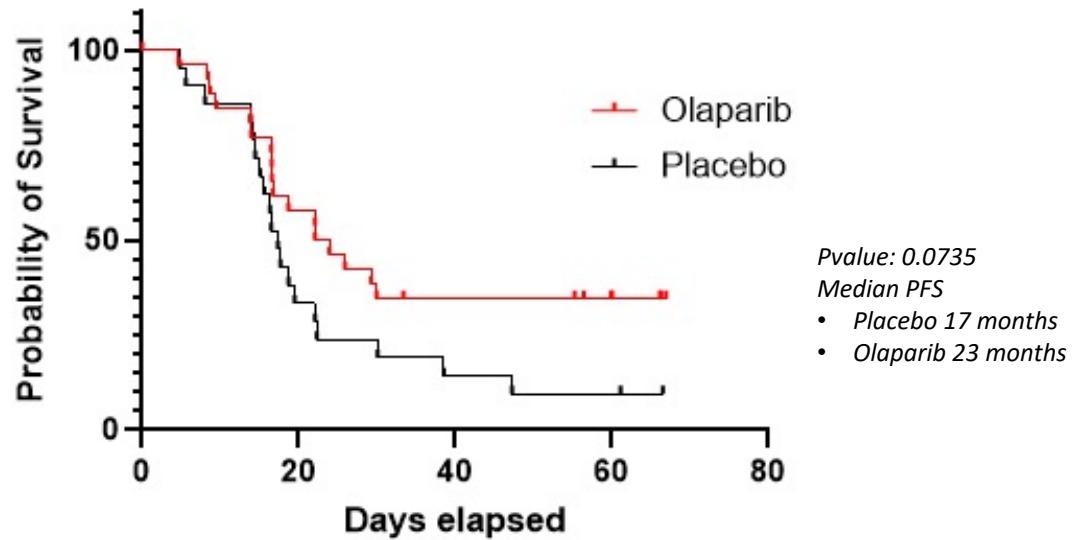
The **HRD test** is a combination of the analysis of the BRCA1/2 status and the genomic scar (GS).

The module is based on the analysis of highly polymorphic SNPs from dbSNP with MAF>0.3 (around 10,000 across the genome). These selected SNPs are distributed along the genome and on telomeric regions . LOH is computed on targeted genes and HRD score is computed on 3 ways:

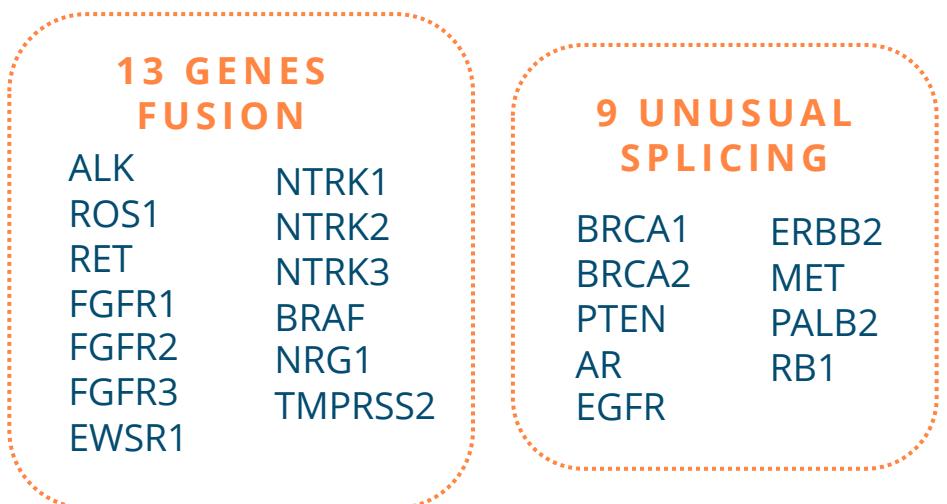
a global score called genomic scar (GS) on all the targets:

- a score only on the Allelic Disparity on Telomere (ADT)
- a score on all the regions except the telomeric ones (LOH)
- a score of large-scale Rearrangements (LR)

GS is considered as positive if > 39



# Design and Content Considerations



22 driver genes but not only.....

Cancers	Targeted kinase fusion partners	Drug	Approval, year	Reference
Ph <sup>+</sup> CML <sup>a</sup>	<i>BCR-ABL1</i>	Imatinib	2001	Amarante-Mendes <i>et al.</i> <sup>75</sup> , Milojkovic and Apperley <sup>76</sup>
Ph <sup>+</sup> CML <sup>a</sup>	<i>BCR-ABL1</i>	Nilotinib	2010	Amarante-Mendes <i>et al.</i> <sup>75</sup> , Radich <i>et al.</i> <sup>77</sup>
NSCLC ALK or <i>ROS1</i> positive <sup>b</sup> , ALCL ALK positive <sup>c</sup>	ALK, <i>ROS1</i> , MET	Crizotinib	2011	Shaw <i>et al.</i> <sup>78</sup> , Solomon <i>et al.</i> <sup>79</sup>
NSCLC ALK positive <sup>d</sup>	ALK, <i>ROS1</i>	Ceritinib	2014	Facchinetto <i>et al.</i> <sup>80</sup>
NSCLC ALK positive <sup>d</sup>	ALK, <i>ROS1</i> , EGFR	Brigatinib	2017	Descourt <i>et al.</i> <sup>81</sup>
NSCLC ALK positive	ALK	Alectinib	2017	Wang <i>et al.</i> <sup>82</sup>
Solid tumors with <i>NTRK</i> gene fusions	<i>NTRK1-3</i>	Larotrectinib	2018	Rudzinski <i>et al.</i> <sup>83</sup>
NSCLC <i>ROS1</i> positive <sup>e</sup> , solid tumors with <i>NTRK</i> fusions	<i>NTRK</i> , <i>ROS1</i> , ALK	Entrectinib	2019	Dziadziuszko <i>et al.</i> <sup>84</sup> , Doebele <i>et al.</i> <sup>85</sup>
Urothelial cancer with <i>FGFR3</i> fusion	<i>FGFR3</i>	Erdafitinib	2019	Zengin <i>et al.</i> <sup>86</sup>
Cholangiocarcinoma with <i>FGFR2</i> fusion	<i>FGFR1-3</i>	Pemigatinib	2020	Abou-Alfa <i>et al.</i> <sup>87</sup>
<i>RET</i> -positive NSCLC, thyroid cancer <sup>f</sup>	<i>RET</i>	Pralsertinib	2020	Gainor <i>et al.</i> <sup>88</sup>
<i>RET</i> -positive NSCLC, thyroid cancer <sup>f</sup>	<i>RET</i> , <i>VEGFR1</i> , <i>VEGFR3</i>	Selpercatinib	2020	Subbiah <i>et al.</i> <sup>89</sup>
NSCLC ALK positive	ALK	Lorlatinib	2021	Sehgal <i>et al.</i> <sup>90</sup> , Descourt <i>et al.</i> <sup>81</sup>
Cholangiocarcinoma with <i>FGFR2</i> fusion	<i>FGFR2</i>	Infigratinib	2021	Javle <i>et al.</i> <sup>91</sup>
Ph <sup>+</sup> CML <sup>a</sup>	<i>BCR-ABL1</i>	Asciminib	2021	Yeung <i>et al.</i> <sup>92</sup>

PMID: 36601633

AGTRAP	BRCA2	DCTN1	ETV5	FGFR3	KIF5B	NPM1	PRKAR1A	ROS1	TMPRSS2
AKAP9	CCDC6	EGFR	ETV6	FN1	KLC1	NRG1	PTEN	SDC4	TPM3
ALK	CD74	EML4	EWSR1	GOLGA5	KTN1	NTRK1	QKI	SLC34A2	TPM4
AR	CEP89	ERBB2	EZR	GOPC	LMNA	NTRK2	RAF1	SLC3A2	TPR
ATIC	CLCN6	ERC1	FAM131B	HIP1	LRIG3	NTRK3	RANBP2	STRN	TRIM24
BRAF	CLIP1	ETV1	FGFR1	HOOK3	MET	PALB2	RB1	TACC3	TRIM27
BRCA1	CLTC	ETV4	FGFR2	KIAA1549	NCOA4	PDGFRB	RET	TFG	TRIM33



# OncoKDO: Our hidden treasure

OncoKDO is our dynamic and proprietary database that is the cornerstone of our intelligence.

## DESIGN: All relevant data matter

→ OncoKDO enables a unique holistic approach



## DATA ACCURACY:

Dynamic and continuously growing database

Since 2013



Manual, daily  
data curation  
by our scientists



### Proprietary input

- Clinical data from 46,000 patients today
- Diagnostic test results
- Raw data upload
- Treatment decisions and outcome, including follow-up



### External sources

- > 15 public databases
- Clinical literature

# Future Directions

ACVR2A	EGF	HIP1	MYB	PRDM10	TMPRSS2	BICC1	FLI1	MSH2	SATB2
AGTRAP	EGFR	HIST1H3B	MYBL1	PRKACA	TP53	BOC	FLT1	MYH9	SDC1
AKAP9	EML4	HMGA2	MYC	PRKACB	TPM3	BTBD1	FLT3	MYO1E	SERPINE1
AKT1	EPC1	HOOK3	MYOD1	PRKAR1A	TPM4	C11orf95	FRMD6	NAB2	SFPQ
AKT2	ERBB2	HRAS	NCOA1	PRKCA	TPR	C8orf42	GLIS1	NACC2	SH3PXD2A
AKT3	ERBB4	IDH1	NCOA2	PRKCB	TRIM11	CARS	GLIS3	NEDD4	SHTN1
ALK	ERC1	IDH2	NCOA3	PRKCD	TRIM24	CCAR2	GPR128	NELFCD	SMAD3
AR	ERG	IGF1R	NCOA4	PRKD1	TRIM27	CCDC88A	GRM1	NFIX	SMARCA2
ARHGAP26	ESR1	INSR	NFATC2	PRKD2	TRIM33	CCND2	HAS2	NONO	SMARCB1
ARHGAP6	ESRRA	JAK2	NFE2L2	PRKD3	USP6	CDC42BPB	HEY1	NOTCH3	SND1
ATIC	ETV1	JAK3	NFIB	PTEN	VGLL2	CDC42EP2	HSPA8	NPEPL1	SNX14
AXL	ETV4	JAZF1	NOTCH1	QKI	WWTR1	CDH11	HTRA1	NRG2	SPRED2
BCOR	ETV5	KEAP1	NOTCH2	RAD51B	YAP1	CDK4	IRF2BP2	NSD2	SQSTM1
BRAF	ETV6	KIAA1549	NPM1	RAF1	YWHAE	CITED2	IRF4	NSD3	SRF
BRCA1	EWSR1	KIF5B	NR4A3	RANBP2	ABL1	CNTRL	KDM2A	NUTM2A	SSX1
BRCA2	EZR	KIT	NRAS	RB1	ACTB	COL12A	KDR	NUTM2B	SSX2
BRD3	FAM131B	KLC1	NRG1	RELA	ACTN4	COL1A1	KIAA1217	NUTM2E	SSX4
BRD4	FGF1	KRAS	NTRK1	RET	AFAP1	COL1A2	KLF17	PAN3	STAT3
CAMTA1	FGFR1	KTN1	NTRK2	ROS1	AGAP3	COL3A1	KMT2A	PATZ1	STX16
CCDC6	FGFR2	LMNA	NTRK3	RSPO2	AGK	COL6A3	LEUTX	PAX7	SUZ12
CCNB3	FGFR3	LRIG3	NUMBL	RSPO3	AHRR	CREB1	LPP	PBX1	SYNCRIP
CCND1	FGR	LTK	NUTM1	SDC4	AK7	CREB2L2	LSM14A	PBX3	TACC1
CD274	FN1	MAML2	PALB2	SLC34A2	AKAP12	CREB3L1	MAF	PML	TACC2
CD74	FOS	MAP2K1	PAX3	SLC3A2	AKAP4	CREB3L2	MAGI3	POLA2	TAF68
CEP89	FOSB	MAP3K3	PAX8	SS18	ASPL	CREBBP	MAML1	POU5F1	TBL1XR1
CIC	FOXO1	MAP3K8	PDGFB	SS18L1	ASPSCR1	DDIT3	MAML3	PPARGC1A	TEAD1
CLCN6	FOXO4	MAST1	PDGFD	STAT6	ATF1	DUX4	MARK2	PPFIA1	TCFP2
CLIP1	FOXR2	MAST2	PDGFRA	STK11	ATM	E1AF	MBNL1	PPP1CB	TGFB3
CLTC	FUS	MBTD1	PDGFRB	STRN	ATP1B1	EIF3K	MEF2A	PRCC	TM30
CRTC1	GLI1	MDM2	PHF1	TACC3	ATRX	EMILIN2	MEIS1	PRKAR1B	TRIO
CSF1	GNA11	MEAF6	PHKB	TAF15	AUTS2	EP400	MGA	PTPRR	VGLL3
CSF1R	GNAQ	MET	PIK3CA	TCF12	BAG4	ERBB2	MGMT	PTPRZ1	VIM
CTNNB1	GNAS	MGEA5	PKN1	TERT	BCL2	ETS1	MIR143	RARA	WT1
CYSLTR2	GOLGA5	MKL2	PLAG1	TFE3	BCL6	EXOC2	MITF	RELCH	ZFP36
DCTN1	GOPC	MN1	POLD1	TFEB	BCLAF1	FBXO25	MKL1	RNF213	ZFTA
DDR2	GRB7	MSMB	POLE	TFG	BCORL1	FEV	MKRN1	RPS6KB1	ZMYM2
DNAJB1	H3F3A	MUSK	PPARG	THADA	BCR	FGFR4	MLLT3	RREB1	ZNF444
									ZNF703

Big RNA genes panel dedicated to solid tumour

- 371 genes for detection of fusions, splicings, exon-skippings
- Fusion genes dedicated to sarcoma diagnosis
- Detection of unknown partner by shadow sequencing

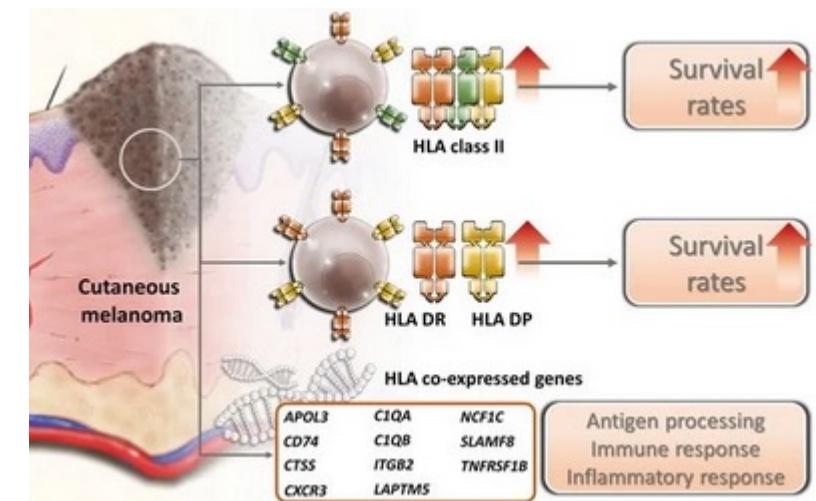
# Future Directions

## APOBEC signature:

- Specific pattern of mutations in DNA, increasing frequency of C to T and G to A
- Identified in various cancer types
- Involved in tumour evolution, potential therapeutic strategies (ICI, Chemotherapy) and prognosis indicator

## HLA signature

- HLA genes already present in the OncoDEEP Kit genes panel
- Specific patterns of HLA gene expression and variation in individuals, which can influence immune responses and has prognosis value in various cancer type
- HLA types associated with better or worse outcomes under immunotherapies



<https://www.mdpi.com/2075-4418/9/2/59>



## European User Group Meeting OncoDEEP® Kit

21<sup>st</sup> - 22<sup>nd</sup> October 2024

# OncoDEEP Kit: The SEGLH Optimisation and Validation Experience

Dr Gareth Gerrard

*Clinical Scientist & Scientific Lead for Cancer Genomics*

South East Genomic Laboratory Hub, Guy's & St. Thomas' NHS Trust  
Synnovis Cancer Genetics, London, UK

# Key SEGLH / Synnovis People for OncoDEEP Validation

Katya  
Mokretar

Lucas Pavlou

Sasha Hansel

Cancer  
Genetics Lab  
Team

Bioinformatics  
Team

# Multistage Optimisation & Validation



## 1. HRD

- To meet needs of Test Directory
- Separate validation cohort



## 2. SNVs

- Small variants
- Indels
- Phased service implementation

## 3. CNV

- Copy number gains
- Copy number losses

## 4. MSI & TMB

- Microsatellite instability
- Tumour Mutational Burden

# Four Types of CNV Calls (including WT call)

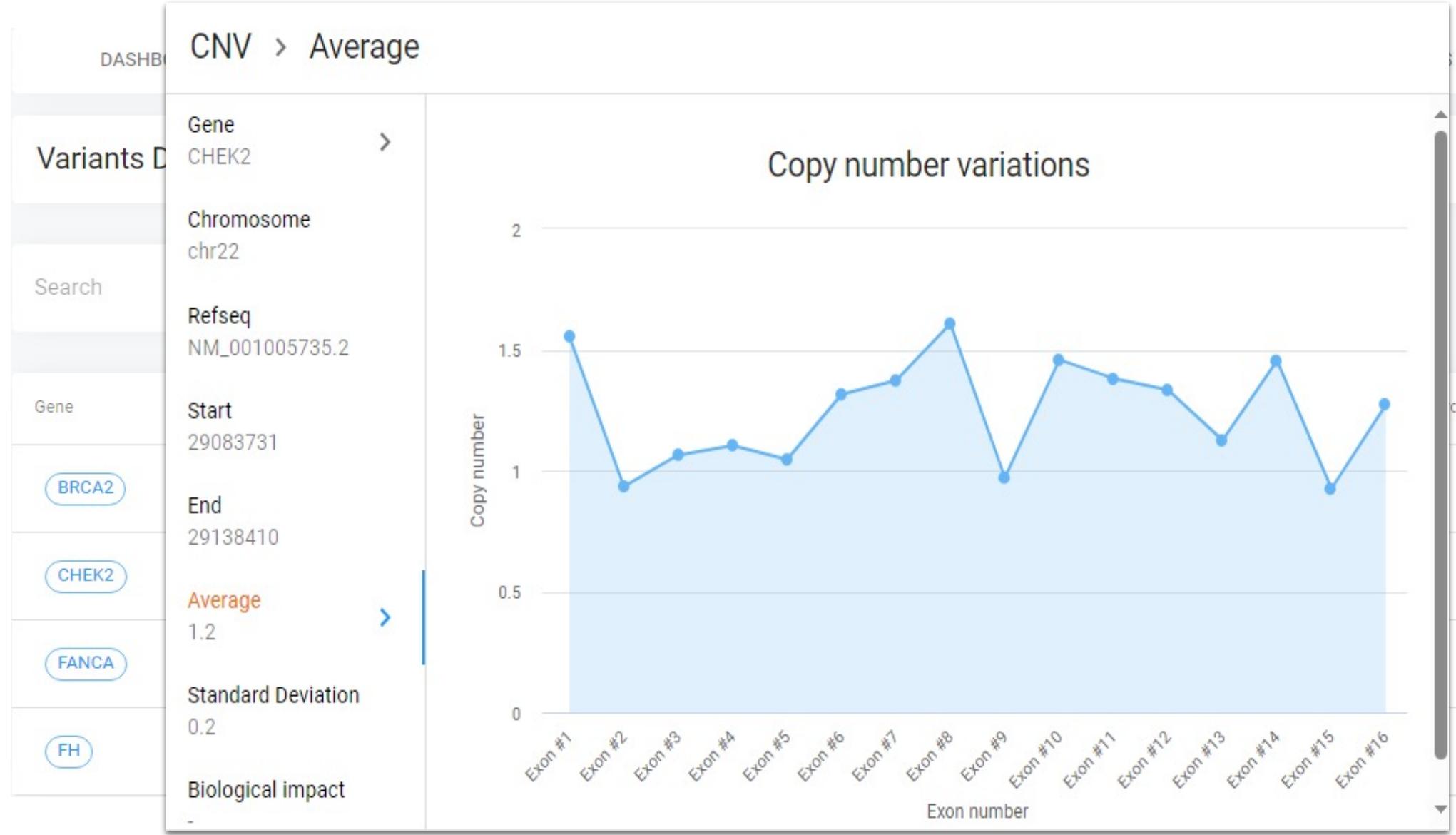
DASHBOARD MEDICAL INFORMATION DRUGS COMPREHENSIVE SUMMARY **VARIANTS DETECTION** OTHER BIOMARKERS CLINICAL TRIALS QUALITY C

Variants Detection > **VARIANTS** ALPHA LIST CNV CNV BY TARGET CNA

Search  Filter by genes  Filter by potential germline variants  All Filter by variant type  All

Gene	Category	Variant Frequency	Copy Number	cDNA Variant	Amino Acid Variant	Impact location	Biological impact
BRCA2	CNV LOSS	0	0	-	-	-	Pathogenic
EGFR	SNV	3.12%	-	NM_005228.5:c.2573T>G	NP_005219.2:p.(Leu858Arg)	-	Pathogenic
CHEK2	LOH	0	1	-	-	-	Likely Pathogenic
FANCA	LOH	0	1	-	-	-	Likely Pathogenic
FH	CNV AMPL	0	6.27	-	-	-	Likely Pathogenic

# Click Through for Granular CNV Details



# CNV Validation Set: 1. Commercial Control Materials

1. Seraseq Breast CNV Mix (+6 copies *ERBB2*, *FGFR3*, *MYC*)
2. Seraseq Lung & Brain CNV Mix (+6 copies *EGFR*, *MYCN*, *MET*)
3. Seraseq Breast CNV Mix (+12 copies *ERBB2*, *FGFR3*, *MYC*)
4. Seraseq Lung & Brain CNV Mix (+12 copies *EGFR*, *MYCN*, *MET*)
5. Horizon Discovery HD200; SNV/Indel control

# 50:50 Mix of SeraCare Breast and Lung & Brain Controls

SeraCare +6 Mix (50:50); Expected Copy Number= 5

1: 232540_232540			2: OKDPAL0003_23_2540_42_aR			3: OKDPAL0003_23_2540_42_a								
	CN	SD	Call		CN	SD	Call		CN	SD	Call	Mean	SD	CV
MET	6.8	1.7AMPL	MET		7.6	2.0AMPL	MET		7.9	2.1AMPL		<b>7.4</b>	0.46	6.2%
EGFR	6.3	1.5AMPL	EGFR		6.3	1.6AMPL	EGFR		6.6	1.7		<b>6.4</b>	0.14	2.2%
MYCN	4.7	1.2	MYCN		4.8	1.5	MYCN		5.0	1.6		<b>4.8</b>	0.12	2.6%
ERBB2	4	0.5	ERBB2		3.9	0.4	ERBB2		4.3	0.5		<b>4.1</b>	0.17	4.2%
MYC	3.9	0.3	MYC		4.0	0.4	MYC		4.2	0.4		<b>4.0</b>	0.12	3.1%
FGFR3	3.5	0.5	FGFR3		3.5	0.6	FGFR3		3.7	0.6		<b>3.6</b>	0.09	2.6%
CDKN2A	1.5	0.5	CDKN2A		1.4	0.4	CDKN2A		1.4	0.4		<b>1.4</b>	0.05	3.3%

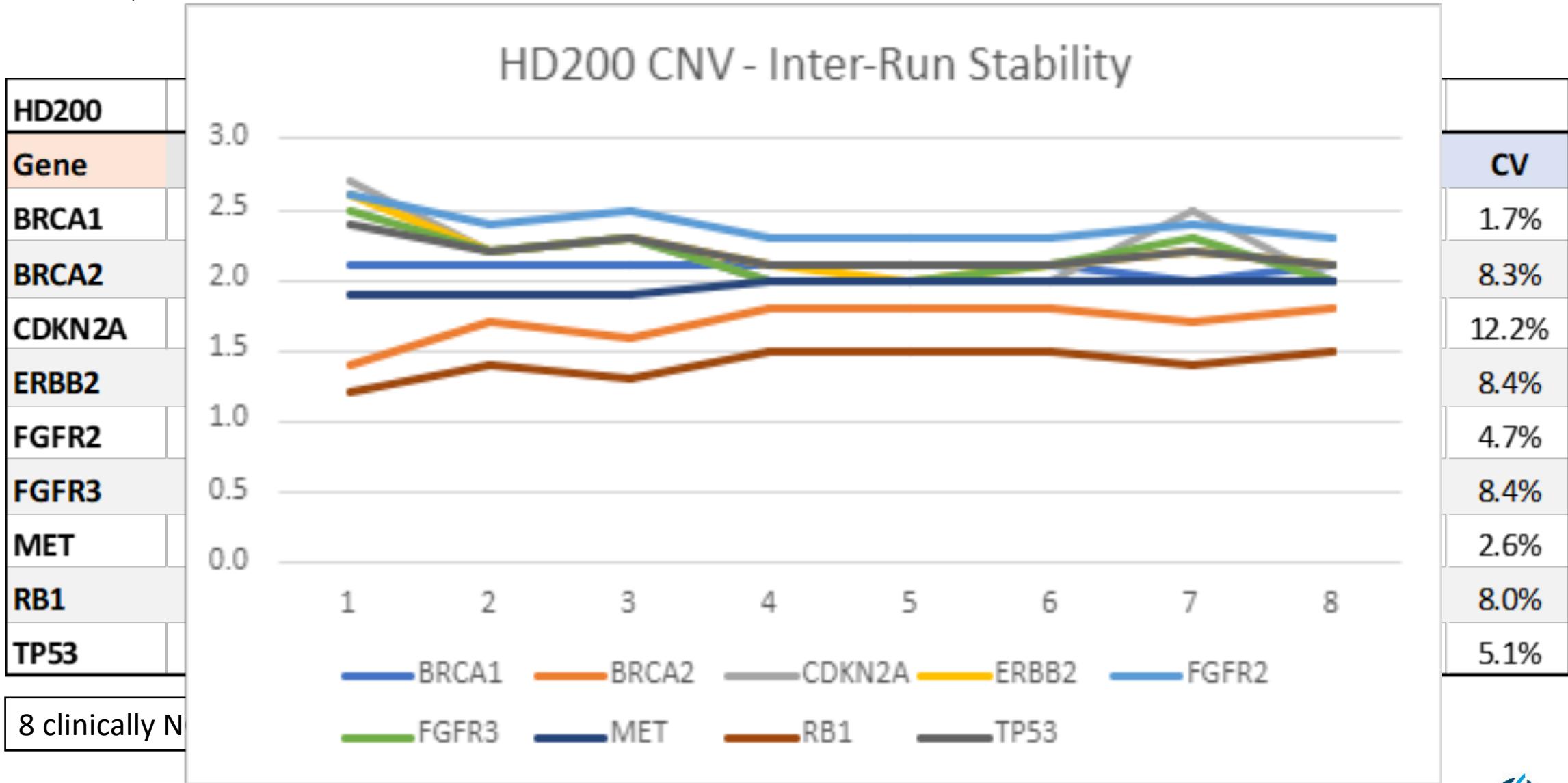
Mean of means= 5

SeraCare +12 Mix (50:50); Expected Copy Number= 8

1: 232541_232541			2: OKDPAL0003_23_2541_43_aR			3: OKDPAL0003_23_2541_43_a								
	CN	SD	Call		CN	SD	Call		CN	SD	Call	Mean	SD	CV
MET	8.3	2.3AMPL	MET		9.4	2.6AMPL	MET		10.1	2.9AMPL		<b>9.3</b>	0.76	8.2%
EGFR	8.1	1.9AMPL	EGFR		9.7	2.4AMPL	EGFR		10.6	2.7AMPL		<b>9.5</b>	1.05	11.1%
MYCN	5.6	1.5	MYCN		6.0	1.9	MYCN		6.3	2.0		<b>6.0</b>	0.29	4.8%
ERBB2	8.4	1.0AMPL	ERBB2		8.1	0.9AMPL	ERBB2		9.2	1.0AMPL		<b>8.6</b>	0.47	5.5%
MYC	7.2	0.7AMPL	MYC		7.8	0.8AMPL	MYC		8.5	0.8AMPL		<b>7.8</b>	0.53	6.8%
FGFR3	6.2	0.9AMPL	FGFR3		6.6	1.1AMPL	FGFR3		7.3	1.2AMPL		<b>6.7</b>	0.45	6.7%
CDKN2A	1.5	0.5	CDKN2A		1.4	0.4	CDKN2A		1.4	0.3		<b>1.4</b>	0.05	3.3%

Mean of means= 8

# Horizon Discovery HD200 SNV IQC Control (2 Batches, 8 Runs)



# CNV Validation Set: 2. Clinical Samples

Tumour Type	n=
NSCLC	14
Prostate	8
Colorectal	6
Thyroid Papillary	4
Bladder	3
Melanoma	3
Breast	2
Cholangiocarcinoma	2
Endometrial	2
GIST	2
CUP	1
Ovarian	1
Pancreatic	1
<b>Tot=</b>	<b>49</b>

	ALL	CN $\geq$ 6	ERBB2 $\geq$ 6	Sub-Analyses	
	TOT	49	49	BRCA	BRCA $\leq$ 1.6
TP	39	12	5	9	9
TN	5	36	3	16	18
FP	0	0	0	3	1
FN	5	1	0	0	0
Sensitivity	88.6%	92.3%	100.0%	100.0%	100.0%
Specificity	100.0%	100.0%	100.0%	84.2%	94.7%
Accuracy	89.8%	98.0%	100.0%	89.3%	96.4%
PPV	100.0%	100.0%	100.0%	75.0%	90.0%
NPV	69.3%	97.6%	100.0%	100.0%	100.0%

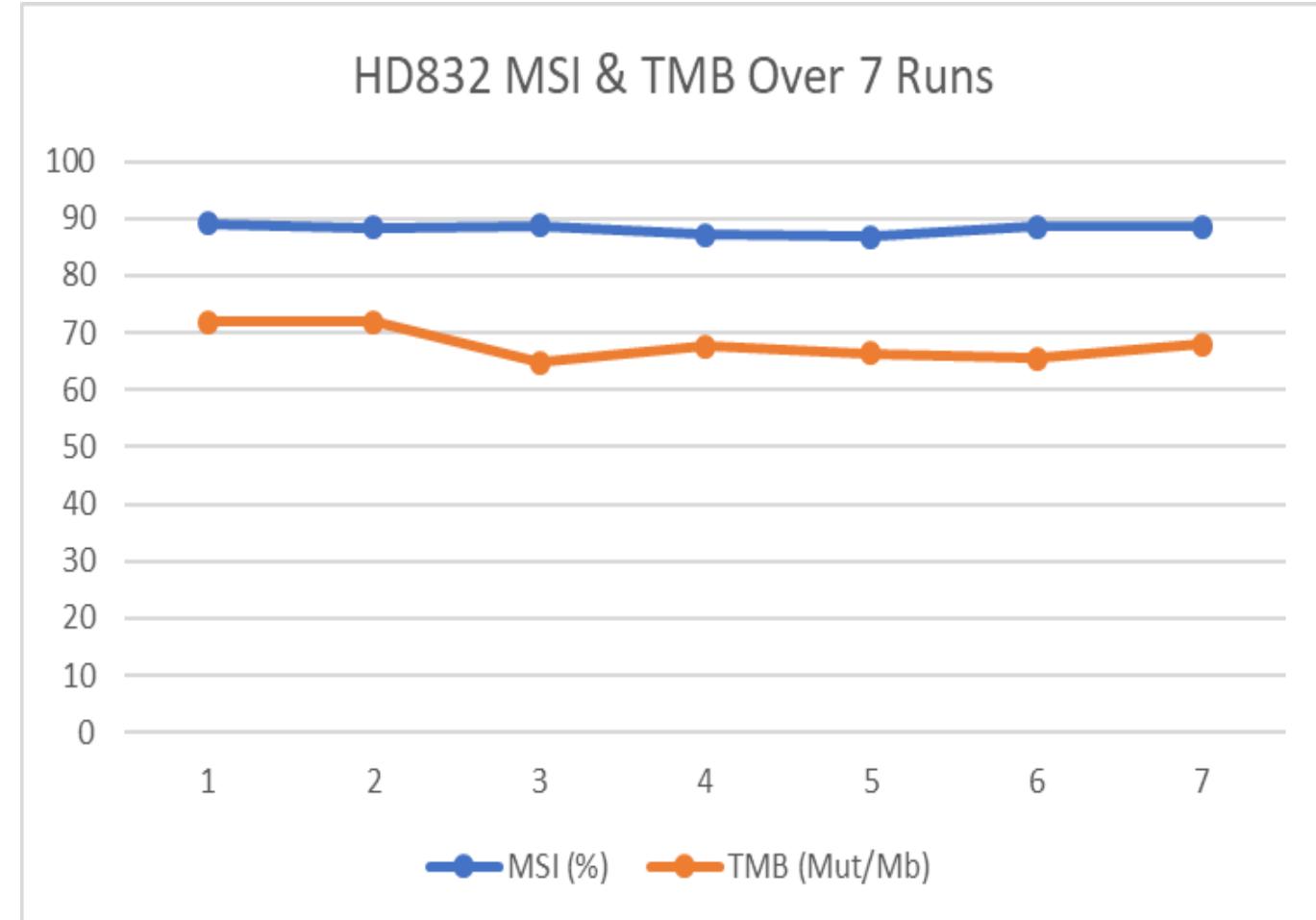
Paired orthogonal results against 2 other SOC NGS panels

# MSI & TMB Validation Set: 1. Commercial Control Materials

1. Horizon Discovery HD832 - OncoSpan
2. Horizon Discovery HD200 – IQC Control
3. SeraSeq MSI Control \*no good for NGS\*
4. SeraSeq TMB Controls \*technical issues\*

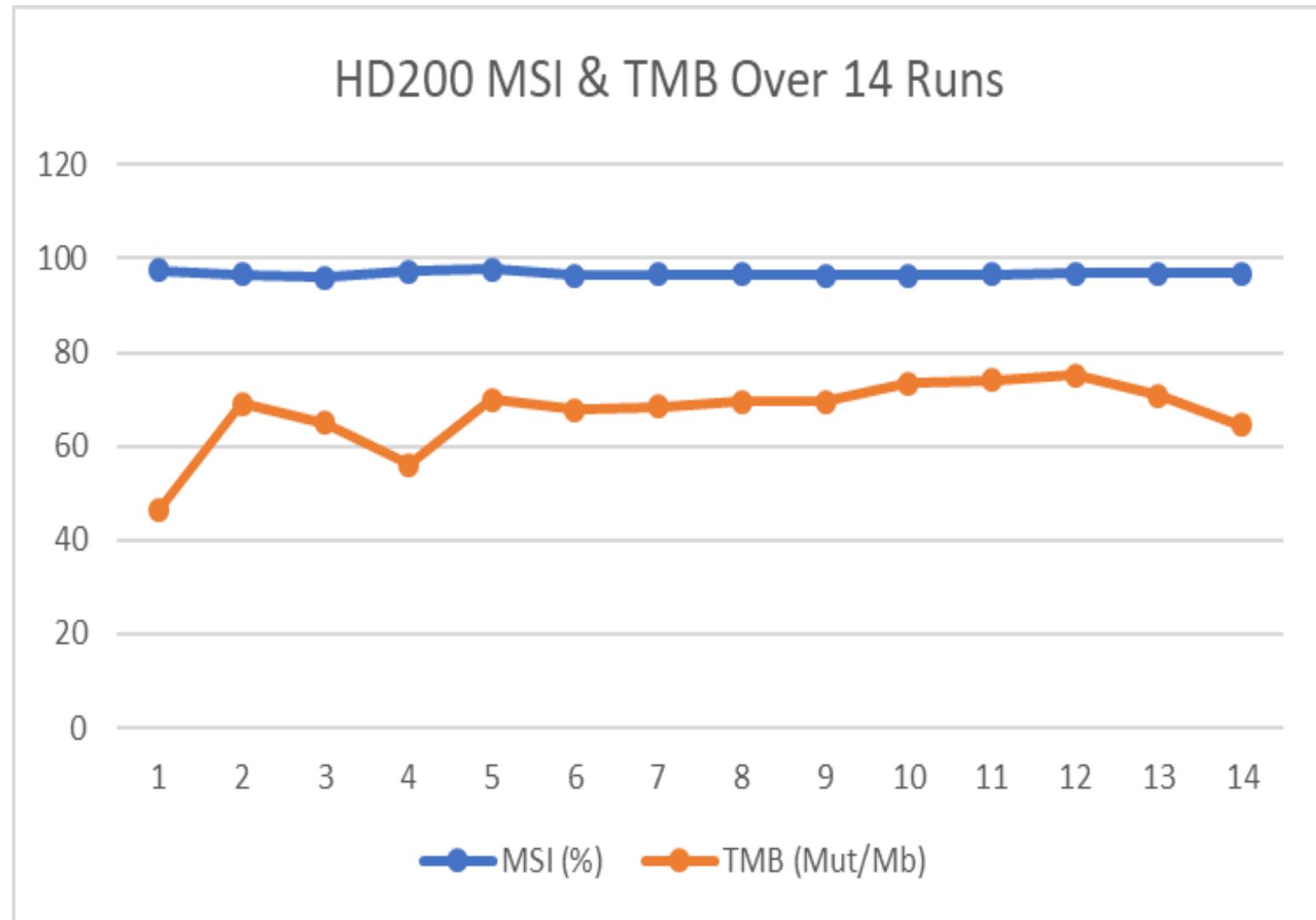
# HD832 – Oncospan – 3 Batches over 7 Runs

HD832 (OncoSpan)			
Run	Batch	MSI (%)	TMB (Mut/Mb)
1	1A	89.01	71.87
2	1B	88.3	71.94
3	2A	88.74	64.7
4	2A	87.09	67.62
5	2B	86.82	66.3
6	3	88.56	65.44
7	3	88.48	67.98
		Mean	88.1
		SD	0.8
		Max	89.0
		Min	86.8
		CV	0.9%
			68.0
			2.7
			71.9
			64.7
			4.0%



# HD200 – IQC – 2 Batches over 14 Runs

HD200			
Run	Batch	MSI (%)	TMB (Mut/Mb)
1	1A	97.42	46.6
2	1B	96.63	68.96
3	2A	95.89	64.96
4	2A	97.3	55.98
5	2A	97.72	69.96
6	1B	96.4	67.86
7	1B	96.66	68.32
8	2A	96.55	69.48
9	2B	96.38	69.46
10	2B	96.47	73.37
11	2B	96.55	73.94
12	2B	96.72	75.06
13	2B	96.8	70.71
14	2B	96.72	64.4
Mean		96.7	67.1
SD		0.5	7.3
Max		97.7	75.1
Min		95.9	46.6
CV		0.5%	10.9%



# MSI – 2. Clinical Orthogonal Validation Set vs SOC NGS

MSI	
Cancer Type	n=
Endometrial	15
CRC	14
Prostate	6
GIST	4
Thyroid	4
Bladder	2
Cholangio	1
CUP	1
Ovarian	1
NSCLC	1

MSI Orthogonal Validation Summary	
TOT	49
TP	15
TN	33
FP	1
FN	0
Sensitivity	100.0%
Specificity	97.1%
Accuracy	98.0%
PPV	93.8%
NPV	100.0%

## TMB – 2. Clinical Orthogonal Validation Set vs SOC NGS

TMB	
Cancer Type	n=
Endometrial	16
CRC	15
Prostate	10
Thyroid	7
GIST	6
Bladder	4
Cholangio	3
CUP	2
Pancreatic	2
Ovarian	1
NSCLC	1
RCC	1
Salivary gland	1

TMB Orthogogonal Validation Summary	
TOT	69
TP	20
TN	41
FP	0
FN	8
Sensitivity	71.0%
Specificity	100.0%
Accuracy	88.0%
PPV	100.0%
NPV	90.0%

# MSI – 3. Clinical Orthogonal vs SOC Fragment Analysis

		OncoDEEP							SOC Fragment Analysis								
		Sample	Result	BAT-25	BAT-26	NR-21	NR-27	D2S123	Sample	Result	BAT-25	BAT-26	NR-21	MONO-27	NR-24		
Sample	Tumor	1	Stable 3.22%	stable	stable	stable	stable	stable	1	MSS	S	S	S	S	S	Score	
1	CRC	2	Stable 2.96%	stable	stable	stable	stable	stable	2	MSS	S	S	S	S	S	TN	
2	CRC	3	Stable 2.34%	stable	stable	stable	stable	failed	3	MSS	S	S	S	S	S	TN	
4	CRC	4	Stable 2.96%	stable	stable	stable	stable	stable	4	MSS	S	S	S	S	S	TN	
5	CRC	5	Unstable 49.13%	stable	unstable	stable	unstable	unstable	5	MSI-H 4/5	US	US	US	US	S	TP	
6	CRC	6	Unstable 70.23%	unstable	unstable	unstable	unstable	unstable	6	MSI-H 5/5	US	US	US	US	US	TP	
7	CRC	7	Unstable 82.74%	unstable	unstable	unstable	unstable	stable	7	MSI-H 5/5	US	US	US	US	US	TN	
8	SCC	8	Stable 22.38%	stable	stable	unstable	stable	stable	8	MSI-L (1/5)	S	S	S	US	S	patient as S10 - our sample	
9	CRC	9	Stable 4.18%	stable	stable	stable	stable	stable	9	MSS	S	S	S	S	S	TN	
10	CRC	10	Stable 2.78%	stable	stable	stable	stable	stable	10							patient as S9 - blood D as 2443268	
11	CRC	11	Unstable 46.39%	stable	stable	stable	stable	stable	11	MSI-H 3/5	US	S	US	US	S	TP	
12	CRC	12	Stable 6.61%	stable	failed	failed	stable	stable	12							patient as S11 - sample	

# Summary

## OncoDEEP Kit now SEGLH large DNA NGS Panel

- Went live for SNVs July 2024
- HRD currently send-away, but coming in-house soon

## CNV Calling is now LIVE!

- October 2024
- CN  $\geq 6$  for gains
- CN  $\leq 1.6$  for losses

## MSI will go live December 2024

- Extra time to allow for team handover

## TMB will also go live

- As 'additional finding' metric