SERVICE PROVISION

The Royal Marsden, together with The Institute of Cancer Research, is the largest comprehensive cancer centre in Europe. This offers a unique environment for cancer research and clinical care, allowing Clinical Genomics to operate as a fully embedded service, benefiting from close interaction with world leading experts in basic and translational research, pathologists & oncologists, across the North Thames Genomic Laboratory Hub. Each request accepted by the laboratory for examination(s) shall be considered an agreement. Requesting, examination and reporting information required for this agreement are documented or linked in this service provision.

Our specialised Clinical Genomics service uses advanced technology to provide a comprehensive range of specialised genomic cancer tests focused on rapid and accurate profiling of solid tumours and haematological malignancies. Our main focus is to provide access to personalised medicine by improving diagnostic strategies, molecular stratification of cancer patients and the suitability of targeted therapies. We offer a responsive and timely service fully staffed by personnel with relevant skills and expertise, with the view to rapidly implement innovative genomic investigations specific to oncology.

Clinical Genomics at the Royal Marsden is a UKAS accredited medical laboratory No. 20653, currently this reflects Molecular Diagnostics examinations and Cytogenetics examinations are accredited as medical laboratory 8444, there is an extension to scope being prepared to merge these two schedules to 20653 with a projected date of February 2022. The schedules are available on the www.ukas.com website on the above reference numbers. Please contact the Business Administrator emma.arton@rmh.nhs.uk for further information

(A) Supplier staff and contact numbers

Clinical Genomics	Role	Telephone Number	Email Address
Main Office / General Enquiries	Molecular Diagnostics	020 8925 6565	Rmh-tr.moleculardiagnostics@nhs.net
	Cytogenetics	020 8722 4232	Rmh-tr.cytogenetics@nhs.net
	Histopathology	020 8915 6648	Rmh-tr.histosspectest@nhs.net
	Central Specimen Reception	020 8915 6570	
Dr Lisa Thompson	Head of Commercial Development	020 8915 6565 Cordless 1006	Lisa.Thompson@rmh.nhs.uk
Julie Howard-Reeves	Operational Lead and Lead Scientist – North Thames GLH and RMH	020 8722 4232	Julie.Howard-Reeves@icr.ac.uk
Dr Mikel Valganon Petrizan	Deputy Operational Lead – RMH	020 8915 6543	mikel.valganonpetrizan@rmh.nhs.uk
Dr Suzanne MacMahon	Senior Clinical Scientist (Solid tumours - Molecular)	020 8915 6587	Suzanne.MacMahon@icr.ac.uk
Dr Jamshid Khorashad	Consultant Clinical Scientist (Haemato-oncology - Molecular lead)	020 8915 6587	Jamshid.Khorashad@icr.ac.uk
Fran Aldridge	Senior Clinical Scientist Cytogenetics	020 8722 4232	Frances.Aldridge@icr.ac.uk
Emma Poyastro-Pearson	Operational Manager – Molecular Diagnostics	020 8915 6561	Emma.Poyastro-Pearson@rmh.nhs.uk
Elisa Garimberti	Operations Manager – Cytogenetics	020 8722 4232	Elisa.Garimberti@rmh.nhs.uk
Marianne Wall	Clinical Scientist (Solid tumour cytogenetics lead)		Marianne.Wall@icr.ac.uk
Tracy Thornton	Clinical Scientist (Haemato-oncology - Cytogenetics lead)		Tracy.Stubbs@icr.ac.uk
Ms Dee Collins & Ms Marion Sullivan	Team Administrators	020 8915 6565	Dee.Collins@icr.ac.uk Marion.Sullivan@nhs.net

Lung IHC Testing			
Laboratory		HHMDS@rmh.nhs.uk	
Medical PA		Rmh-tr.histosspectest@nhs.net Medsec.haematopathology@rmh.nhs.uk Tel: 020 8915 6648 Fax: 020 8915 6566	
For lung pathway communication		For PDL1/ALK1 and ROS1: Rmh-tr.histosspectest@nhs.net For EGFR and other lung panel tests: Rmh-tr.moleculardiagnostics@nhs.net	

Type of Query	Contact
Formal and long-term change to service provided	Julie Howard-Reeves (Operational Lead and Lead Scientist - North Thames GLH/RMH) or Mikel Valganon Petrizan (Deputy Operational Lead)
Ad hoc change to gene reporting –please note **in all cases** this must be made in the form of a written request to the clinical scientist with whom the request has been discussed The request will be replied to with any impacts on TATs clearly identified and agreed before any changes made. Heads of service should be included in any email conversations for audit purposes.	**Molecular diagnostics:** Suzanne.MacMahon@icr.ac.uk and rmh-tr.moleculardiagnostics@nhs.net Tel: 020 8915 6587 **Cytogenetics:** Frances.Aldridge@icr.ac.uk Rmh-tr.cytogenetics@nhs.net Tel: 020 8722 4232
Interpretation of clinical results	**Molecular diagnostics:** Suzanne.MacMahon@icr.ac.uk Tel: 020 8915 6521 Jamshid.Khorashad@icr.ac.uk Tel: 020 8915 6587 **Cytogenetics:** Frances.Aldridge@icr.ac.uk Rmh-tr.cytogenetics@nhs.net Tel: 020 8722 4232
Clinical Advice	**SIHMDS: Consultant Haematologist & SIHMDS Clinical Service Lead:** David.Taussig@rmh.nhs.uk / David.Taussig@nhs.net **Solid Tumour: Consultant Medical Oncologist & Clinical Director (Clinical Genomics):** Angela.George@rmh.nhs.uk / Angela.George6@nhs.net
Change in sample status from standard to urgent priority	**Molecular diagnostics:** Rmh-tr.moleculardiagnostics@nhs.net Tel: 020 8915 6536 **Histopathology:** Rmh-tr.histosspectest@nhs.net Tel: 020 8915 6648

General service enquiries	**Team Administrators** Tel: 020 8915 6565 Tel: 020 8915 6648
O&V System Administrator	Henry.Coleman@rmh.nhs.uk Tel: 07971 913 012 Admin.orderview@rmh.nhs.uk
O&V System Support	Emma.Arton@rmh.nhs.uk Tel: 07736 301 698 Admin.orderview@rmh.nhs.uk
Service Complaints	**Operational lead and Lead Scientist – North Thames GLH & RMH:** Julie.Howard-Reeves@icr.ac.uk **Pathology Quality Team** Pathologyqualityteam@rmh.nhs.uk

(B) Test Repertoire

Clinical Genomics solid tumour sites	
Adrenal	Neurological
Bile Duct	Ovarian
Bladder	Pancreatic
Breast	Prostate
Colorectal	Renal
Gastrointestinal	Salivary Gland
Haematological	Sarcoma
Head & Neck	Skin / Melanoma
Liver	Thyroid
Lung / Mesothelioma	

SIHMDS Molecular Diagnostics
B Cell Clonality
T Cell Clonality
Chimerism
BCR-ABL
NGS panels – please refer to NHSE Cancer Test Directory for details
TP53 gene targeted NGS (CLL)
BRAF gene targeted NGS (HCL)
Translocation panel AML/ALL (RT-PCR)
IGHV Mutational Status
Extended NGS panels – please contact the laboratory for further details
SIHMDS Cytogenetics
Karyotype
FISH

- For full test directory and panels please see NHSE test directory: NHS England
- Please note that additional genes not currently covered by the NHSE test directory are available and can be reported at an additional cost. Please contact the laboratory for further information.
- For details of the genes included in the RMH NGS panels for solid tumours and haematological malignancies, including NTRK fusions please see **Appendix A** below.

(C) Sample Requirements

It is assumed that appropriate patient consent has been obtained by the referral centre.

Solid Tumour	*FISH*	*Tissue*	*Extracted DNA & RNA*
Lung	An FFPE section cut to 1-2µm thickness and mounted on positively charged microscope slides along with an H&E slide with the area of interest clearly marked. Two FFPE slides are required for each FISH probe required to allow the option of repeating the FISH study should hybridisation fail. ***We currently only do FISH for lung following an equivocal IHC result.***	**FFPE sections** - DNA NGS; 5x 10µm unstained slides (uncharged) and RNA NGS - 5x 10µm unstained slides (uncharged). In the case of biopsies 6x 10µm (total for DNA and RNA NGS) unstained slides (uncharged) will be accepted, although this may limit the testing provided. If IHC is required please add an additional two H&E sections and two 1-2 µm mounted sections (unstained, unbaked, positively charged) per test (ALK, PD-L1 and ROS1 IHC testing as required) **(not blocks).** IHC testing is provided by the RMH Histopathology department and separate reports will be issued for the tests performed. Stained and unstained slides are accepted from cytology samples for molecular testing in lung cancer in line with the guidelines from 2018 from AMP, CAP and IASLC	**DNA** - >50 ng extracted DNA per sample, quantified and resuspended in water/buffer at > 1.67 ng/ul. Please specify if DNA is resuspended in water, ATE, EB or EA buffer. **RNA –** 100ng extracted RNA, quantified with a fluorometric assay
Lung ctDNA		Peripheral blood must be collected into cell free DNA collection tubes, ensuring the stabilisation tube is filled to the line. The tube must be inverted 8-10 times following venepuncture & dispatched to the laboratory within 24 hours of sample collection at ambient temperature	
Colorectal *Melanoma* *H&N/Thyroid* *Ovarian* *Glioma* *Sarcoma* *GIST* *Breast* *Other*	*FISH tests for other tumour types would require appropriate validation*	**FFPE sections:** DNA NGS; 5x 10µm unstained slides (uncharged) and RNA NGS - 5x 10µm unstained slides (uncharged). In the case of biopsies 6x 10µm (total for DNA and RNA NGS) unstained slides (uncharged) will be accepted, although this may limit the testing provided.	

SIHMDS	*Sample requirement*
Peripheral Blood (PB)	Cytogenetics studies: • Please send 3-5 ml of PB in Heparin. Molecular studies: • Please send 3-10ml of PB in EDTA except in the case of MRD and Chimerism studies where 10-20ml of blood is recommended. • Samples should be sent on the day of collection and at ambient temperature. Clotted samples are unsuitable for molecular analysis, in these situations please obtain a new sample if possible. • Samples for MRD monitoring should ideally be received 24-48hrs after collection to ensure a reliable result can be obtained.
Bone Marrow (BM)	Cytogenetics studies: • For karyotype studies please send at least 3-5ml of BM in Heparin unless this is not feasible. • For myeloma studies please send 4ml in EDTA Molecular studies: • Please send at least 1ml of BM in EDTA unless this is not feasible (e.g. paediatric samples).
Lymph Nodes / Other tissues	• Fresh tissue in saline • Tissue in fixative • Paraffin embedded tissue, slides scrolls as appropriate • Body fluids can also be processed for Molecular studies if the cell count is at least 10^{5-6}.
Body Fluids	• Cerebrospinal fluid (CSF), Ascitic Fluid (AF), Pleural Fluid (PF) should all be sent fresh and arrive in the laboratory **within 24 hours as cells in these fluids are labile**

Returning material

Unused FFPE slides will be discarded after 6 months, unless prior agreement with histopathology laboratory. The H&E slides will be kept indefinitely.

(D) How to send samples

Transportation and packaging of samples should be performed according to the sender's policy for safe transport of pathological specimens.
See HSE guidance on HSE.Gov

Courier Deliveries:
Preference is for samples to be delivered via Courier.
Clinical Genomics Department
The Centre for Molecular Pathology
The Royal Marsden NHS Foundation Trust
15 Cotswold Road
Sutton, Surrey
SM2 5NG.

Postal Address:
Please note change of postal code if sending via 1st class post:
Clinical Genomics Department
The Centre for Molecular Pathology
The Royal Marsden NHS Foundation Trust
15 Cotswold Road
Sutton, Surrey
SM2 5PT.

Operating Hours:

Central Specimen Reception operating hours: 09:00 – 17:30 Monday to Friday.
Samples received after 15:00 will be processed in the laboratory the following day.

Order & View Requests:

The RMH Order & View portal is a web-based portal that allows users to order tests, track samples and view results online. Email alerts are sent out when results become available.

Access & training for this system is delivered by the Order & View system administrators with support from the RMH system administrator.

For further details please contact the O&V system administrator; Henry.Coleman@rmh.nhs.uk / Admin.OrderView@rmh.nhs.uk or 07971 913 012.

Non-Order & View Requests:

Requests for studies should be made using the appropriate request form.

Request forms can be found on our website here

For enquiries please contact the Business Administrator; Emma.Arton@rmh.nhs.uk or 07736 301 698.

Minimum Data Set:
Please provide the following on all requests:

Patient Demographics	Clinical Details	Sample Details	Treatment Response / MRD	Referrer Details
Patient forename & surname Date of Birth Gender Hospital Number and/or NHS Number **It is the referring organisation's responsibility to ensure the correct identity of the patient and that it corresponds to the information on the request form at the time the sample is taken.** **The sample must have at least two forms of ID that match the ID on the request form.** In the case of Formalin-Fixed Paraffin-Embedded (FFPE) sections and blocks, the histopathology number must be clearly displayed.	Diagnosis: Minimum: - Presentation features, - Hb, - WBC, - Platelets - Stage PLUS: According to the suspected diagnosis: - WBC differential, - Organomegaly, - LDH, - B12/folate status, - Paraprotein type and levels	Date sample taken Sample/tissue type (biopsy/resection) Solid Tumour: - Tumour type, - Specimen type, - Cellularity, - Neoplastic content, - Necrosis, - Increased pigmentation. - Histopathology number	SIHMDS: - Details of treatment including type and duration - Relapse - Prior treatment response	Requesting clinician contact details Email address for report receipt and return of blocks (NHS.Net address is preferred).

Please also include NHS/PP and Urgency status on requests.

(E) Policy for Deviating Samples

In line with TPS 63: UKAS policy on Deviating Samples, the following would be considered 'deviating samples':

- Insufficient tissue and/or tumour (including sample quality, preparation and percentage tumour infiltration)
- Insufficient liquid sample for tests requested
- Inadequate sample labelling / Discrepancies between sample label and request form details
- Inadequate request data quality
- Interference factors e.g. necrosis, melanin (FFPE)
- Inappropriate upstream processing or storage of the sample (FFPE)
- Incorrect collection tube used i.e. anticoagulant
- Unlabelled samples will NOT be accepted.

When a sample has been identified as deviating as defined above, the laboratory, wherever possible will carry out analysis and caveat the report in relation to the sample received.

(F) Report Format

Patient demographics are included on all reports such as full name, date of birth, hospital number and NHS number if available. The laboratory accession numbers are included to ensure that the report unequivocally links to the specific patient and sample sent to the laboratory. The sample type and date of receipt in the laboratory are also indicated.

Technical details, including the tests performed, their limitations and sensitivity are included. Results are presented in a tabulated form, with reference to the genes studied, the scope covered, method used, and in the case of NGS; genotyping results and the transcript reference sequences used to annotate the variants. The interpretation of the results, taking into account the clinical context, is provided as a summary.

If the laboratory has deviated from the service specification described and referenced herein such that it would impact on the examination result, we will inform the referral laboratory.

Please contact the laboratory if further information is required.

(G) Transmission of Reports

	Solid Tumour	*SIHMDS*
Order & View	Results will be available in real time when they are signed out by the individual laboratory.	Where an integrated report (IR) is required test results will be available once the IR is signed out; this is in line with NICE guidance. If results are urgent the laboratories are able to release interim reports ahead of the IR upon request. Where an integrated report is not required the individual reports will be available in real time when they are signed out by the individual laboratories.
Non-Order & View	Individual laboratory reports will be emailed via [secure] NHS.Net mail.	Individual laboratory reports will be emailed via [secure] NHS.Net mail. Integrated Reports, if required, will be sent via [secure] NHS.Net mail once they are available.

Reports normally contain an interpretation of the result. If further advice or interpretation is required, please contact one of the senior staff in the laboratory (see contacts table above). Should a test be performed at an external organisation, this will be stated in the final report.

(H) Quality and Key Performance Indicators

Turnaround Times

Test	*Expected Turn-around Time*
SIHMDS:	
B Cell Clonality	90% of results expected within 14 calendar days
T Cell Clonality	
Chimerism	
BCR-ABL	
TP53 gene targeted NGS (CLL)	
BRAF gene targeted NGS (HCL)	
Translocation panel AML/ALL (RT-PCR)	
NGS panels	
Extended NGS panels (please contact the laboratory for further details)	90% of results expected in 21 calendar days
IGHV Mutational Status	
Cytogenetic FISH and/ or karyotype analysis	**Urgent samples:** within 14 calendar days in 90% of cases **Non-urgent samples:** within 21 calendar days in 90% of cases
Integrated Report	80% of cases in 17 working days

Solid Tumour:*	
Urgent NGS panels (e.g. lung)	90% of results expected within 10-12 calendar days
DNA NGS panel	90% of results expected within 14 calendar days
RNA Fusion panel (including NTRK fusions)	
EGFR – ctDNA (Non-Small-Cell Lung Carcinoma [NSCLC])	
Ovarian cancer NGS panel tBRCA1/2 (ovarian cancer only)	90% of results expected within 21 calendar days
Extended NGS panel (please contact the laboratory for further details)	
Soft tissue sarcoma fusions by FISH (Salvage pathway. Please contact the laboratory for further details)	**Urgent samples:** within 14 calendar days in 95% of cases **Non-urgent samples:** within 21 calendar days in 95% of cases

Performance

All up-to-date quality metrics for SIHMDS and Solid Tumour are available upon request or on the NHS Futures Website

Service Review Process

A quarterly report is available on Key Performance Indicators (including EQA performance), along with assessment of user feedback from both parties.

Complaints procedure

Any complaints concerning the performance of the service should be directed to the Operational Lead and the Pathology Quality Team in the first instance (contact details above).

Any complaints which affect patient care will be dealt with according to the Royal Marsden NHS Foundation Trust's internal incident reporting procedure (please click here for further details).

External Quality Assurance

The laboratories participate in the National External Quality Assurance Schemes which is designed to test the accuracy and performance of all tests within our test repertoire. Where EQA schemes are not available, or the laboratory feels that further assurance is needed they have devised alternative approaches to quality assurance as needed.

For complete details of EQA schemes and any further information please contact the Pathology Quality Team.

Appendix A

RMH Panels Gene lists

RMH200 Version 2 panel

This custom capture panel is property of the Royal Marsden NHS trust and has been clinically validated to cover the following regions as part of the RMH200v2 panel. Analysis is performed using DNA capture and targeted sequencing using the RMH200 panel. Detection of variants is dependent on the percentage of tumour infiltration, DNA input concentration and DNA quality. This panel is capable of detecting gene amplifications (>2 copies) and deletions (<0.5 copies). Structural variant analysis is undergoing validation and calls are advisory only. Where no paired germline control is available, variants of germline origin cannot be excluded. Analysis is performed based on genome build hg19.

Please note that some rare mutations in the genes analysed may not be detected. Alterations outside the specified regions analysed cannot be detected. The limit of detection of these methods needs to be considered together with the level of tumour involvement and the quantity and quality. Some mutations may be present below the level of detection.

ABL1	CCNE1	FANCI	KRAS	PDCD1LG2	SETD2
ACVR1	CCNE2	FANCL	LIN28B	PDGFRA	SF3B1
AKT1	CD79B	FAT1	LZTR1	PHOX2B	SH2B3
AKT2	CDH1	FBXW7	MAP2K1	PIK3CA	SMAD2
AKT3	CDK12	FGF10	MAP2K2	PIK3CD	SMAD3
ALK	CDK2	FGFR1	MAP2K4	PIK3R1	SMAD4
AMER1	CDK4	FGFR2	MAP3K1	PIN1	SMARCA4
ANTRX2	CDK6	FGFR3	MAPK1	PMS1	SMARCB1
APC	CDKN1A	FGFR4	MCL1	PMS2	SMARCE1
AR	CDKN1B	FH	MDM2	POLD1	SMO
ARAF	CDKN2A	FLT3	MDM4	POLE	SOX2
ARID1A	CDKN2B	FOXL2	MEN1	POT1	SRSF2
ARID1B	CDKN2C	FOXO1	MET	PPM1D	STAG2

ARID2	CEBPA	GATA1	MLH1	PPP2R2A	STK11
ASXL1	CHEK1	GATA3	MN1	PRKAR1A	SUFU
ATM	CHEK2	GNA11	MPL	PTCH1	TCEB1
ATR	CIC	GNAQ	MRE11A	PTCH2	TCF3
ATRX	CKS1B	GNAS	MSH2	PTEN	TERT
AURKA	CREBBP	GPR161	MSH6	PTPN11	TET2
AXIN1	CRLF1	H3F3A	MTOR	RAD21	TFE3
AXIN2	CTNNB1	HIST1H3B	MYC	RAD50	TP53
B2M	CXCR4	HIST1H3C	MYCL	RAD51B	TP63
BAP1	DAXX	HIST2H3A	MYCN	RAD51C	TSC1
BARD1	DDR2	HIST2H3C	MYD88	RAD51D	TSC2
BBC3	DDX3X	HRAS	NF1	RAD54L	U2AF1
BCL2	DICER1	IDH1	NF2	RAF1	VHL
BCOR	DROSHA	IDH2	NFE2	RB1	WT1
BCORL1	EGFR	IGF1R	NFE2L2	RBM10	YAP1
BIRC3	ELP1	IRF4	NOTCH1	RET	DPYD
BRAF	EMSY	IRS2	NOTCH2	RhoA	EBF1
BRCA1	EP300	JAK2	NOTCH3	RICTOR	IL3RA
BRCA2	EPHB2	KBTBD4	NPM1	RIT1	KIAA1549
BRIP1	ERBB2	KDM6A	NRAS	RNF43	MYOD1
BTG1	ERBB3	KDR	NTRK1	ROS1	TG
BTK	ESR1	KEAP1	NTRK2	RUNX1	YES1
CALR	ETV6	KIT	NTRK3	SDHA	YWHAE
CASP8	EZH2	KLF2	OTX1	SDHB	H3F3B
CBL	F2R	KMT2A	PALB2	SDHC	ID3

CCND1	FADD	KMT2C	PAX5	SDHD	NA
CCND2	FAM46C	KMT2D	PBRM1	SETBP1	NA

HaemOnc panel

This custom capture panel is property of the Royal Marsden NHS trust and has been clinically validated to cover the following regions as part of the HaemOnc panel. Analysis is performed using DNA capture and targeted sequencing using the HaemOnc panel. Detection of variants is dependent on the percentage of tumour infiltration, DNA input concentration and DNA quality. Where no paired germline control is available, variants of germline origin cannot be excluded. Analysis is performed based on genome build hg19.

Please note that some rare mutations in the genes analysed may not be detected. Alterations outside the specified regions analysed cannot be detected. The limit of detection of these methods needs to be considered together with the level of tumour involvement and the quantity and quality. Some mutations may be present below the level of detection.

ABCA1	CCND3	ERBB3	KLF2	NTRK2	SAMD9L
ABL1	CD274	ETNK1	KMT2A	NTRK3	SAMHD1
ACD	CD79B	ETV6	KMT2C	NUDT15	SBDS
AKT1	CDKN2A	EZH2	KMT2D	NUP98	SETBP1
ANKRD26	CDKN2B	FAM46C	KMT2E	PALB2	SF1
ARAF	CDKN2C	FANCL	KRAS	PAX5	SF3B1
ARID1A	CEBPA	FBXW7	LAMB4	PDGFRA	SH2B3
ASXL1	CHEK2	FLT3	LMO1	PDS5B	SMC1A
ATM	CKS1B	FOXO1	LMO2	PHF6	SMC3
ATR	CREBBP	G6PC3	LUC7L2	PIGA	SRSF2
ATRX	CRLF2	GATA1	MAP2K1	PIK3CA	STAG2
BCL11B	CSF2RA	GATA2	MAP3K1	PIK3CD	STAT3
BCL2	CSF3R	GFI1	MECOM	PIK3R1	STAT5B
BCL2	CSNK1A1	GNAS	MEF2B	PLCG2	SUZ12
BCL6	CTC1	GNB1	MET	PMS1	TAL1

BCOR	CTCF	GPRC5A	MPL	PMS2	TERC
BCORL1	CUX1	HAX1	MYB	PPM1D	TERT
BCR	CXCR4	HRAS	MYC	PRPF8	TET1
BIRC3	DCLRE1C	IDH1	MYCN	PTEN	TET2
BOD1L1	DDX41	IDH2	MYD88	PTPN11	TINF2
BRAF	DIS3	IKZF1	NCOR1	PTPRT	TP53
BRCA1	DKC1	IL3RA	NCOR2	RAD21	TPMT
BRCA2	DNAJC21	IL7R	NF1	RAD50	TRAF2
BRCC3	DNM2	IRF1	NFE2	RAD51C	U2AF1
BTG1	DNMT3A	IRF4	NOTCH1	RAD51D	U2AF2
BTK	DNMT3B	JAGN1	NOTCH2	RB1	USP7
CALR	DUSP22	JAK1	NPM1	RHOA	VPS45
CARD11	DYPD	JAK2	NRAS	RIT1	WAS
CBL	EBF1	JAK3	NRD1	RPL10	WT1
CBLB	EED	KDM5A	NSD1	RPL5	XPO1
CCND1	ELANE	KDM6A	NT5C2	RUNX1	XRCC2
CCND2	EP300	KIT	NTRK1	SAMD9	ZRSR2

RNA Pan-Caner panel

The Illumina Pan Cancer Gene fusion panel targets 1,385 cancer-associated genes and 21,043 exons.

This target enrichment design consist of coding regions including 160 bp of the 5' and 3' UTR of every targeted gene, to ensure full gene coverage. Enrichment was performed with 21,283 probes targeting 1385 genes associated with gene fusions, following manufacturer's protocols. Bioinformatic analysis was performed using the RNA-Seq Alignment App v2.0.1 (BaseSpace Sequencing Hub) using STAR aligner (to RefSeq Homo Sapiens/ hg19 genome) and Manta for gene fusion calling with default parameters.

High confidence fusion calls have a fusion score of >0.6 and ≥ 3 fusion supporting reads. Detection of fusions is currently undergoing validation, therefore any variants should be subject to confirmation by an orthogonal method. The method is not currently suitable for detection of STIL-TAL1 fusions.

The sensitivity of this assay is 100% (95%CI: 66.37% to 100%) with a specificity of 100% (95%:78.20% to 100%); The level of detection (LOD) is under validation. Detection of fusions is dependent on the percentage of tumour infiltration, RNA input concentration and RNA quality. Some fusions may be present below the level of detection, depending on the level of tumour infiltration and the size of the sample sent.