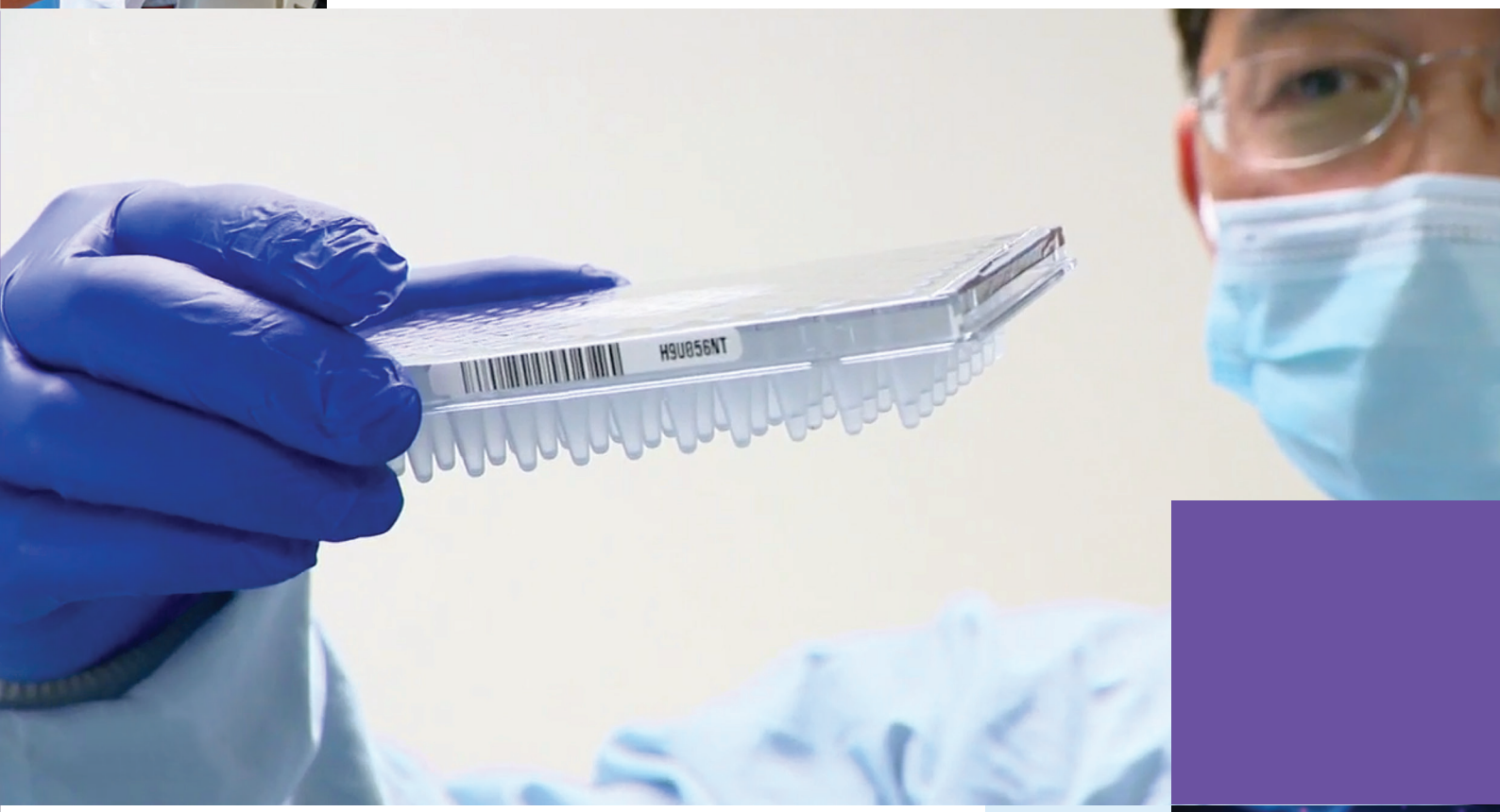
SydPath
ST VINCENT'S PATHOLOGY



SOMATIC MUTATION TESTING

Next Generation Sequencing

www.sydpath.com.au

SydPath provides an unwavering commitment to delivering high quality testing with a fast time to results, combined with our extensive experience to propel us to the forefront of diagnostic pathology, research and scientific development. Our patient centric service is powered by St Vincent's Health Australia, underpinned by advanced scientific pathology and laboratory technology, to optimise outcomes and care. As a teaching hospital laboratory located within the St Vincent's Hospital Campus, SydPath also provides specialised services tailored to the needs of the pharmaceutical and biotechnology industries for their clinical trial and research programs.

SydPath is purpose driven pathology.

SydPath's Departments of Anatomical Pathology, Haematology and Molecular Pathology are pleased to offer comprehensive somatic mutation testing tailored to meet the clinical needs of oncologists, haematologists, surgeons and other clinicians. Somatic genomic testing has now become the standard of care in a variety of cancers for:

- Diagnostic refinement
- Risk stratification
- Therapeutic approaches

As each tumour is different, SydPath delivers an optimised and personalised approach:

- Guaranteed fast turnaround time for results afforded by in-house NGS testing capabilities at our NATA accredited Darlinghurst laboratory.
- Identification of precision oncology treatments based on your patients' tumour molecular profile.
- Every single report undergoes clinical interpretation by a pathologist that holds NPAAC accreditation, ensuring the highest standard of patient care.

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COMPREHENSIVE NEXT GENERATION SEQUENCING

Next generation sequencing (NGS) is the keystone of somatic mutation testing here at SydPath. We currently perform testing on:

- Formalin-fixed paraffin embedded tissue blocks (including cell blocks)
- Cytology samples
- Peripheral blood and bone marrow specimens for haematological indications.

A pathologist conducts thorough quality checks on all samples to ensure their suitability and maintains a high standard of testing.

SydPath is rapidly expanding its NGS and cancer genetics services, offering a range of gene panels on both DNA (for single nucleotide variants, insertions, deletions and copy number variations) and RNA (for gene fusions and rearrangements) .

These panels are applicable to a range of malignant tumour types including:

Lung carcinoma
Colorectal carcinoma
Melanoma
Prostatic carcinoma
Thyroid carcinoma
Breast carcinoma
Myeloid malignancies including acute myeloid leukaemia, myelodysplastic syndromes and myeloproliferative neoplasms
Lymphoid malignancies (currently being validated)
Others including sarcomas

SydPath is New South Wales' leading NGS provider and our service provides a significant step forward in personalised and precision medicine, enabling the detection of somatic driver mutations, resistance mechanisms and quantification of mutational burden.

NGS is a key clinical tool used in comprehensive cancer care in St Vincent's Hospital Sydney. Our aim is to drive awareness and access to this transformative technology, to optimise care and outcomes across New South Wales and beyond. Our current referral base is expansive, including over 20 public and private pathology providers.

SydPath P +61 2 8382 9100 **W** sydpath.com.au

MEET OUR HIGHLY TRAINED & EXPERIENCED SYDPATH TEAM

Our Pathologists

Associate Professor Min Ru Qiu

Senior Molecular Anatomical Pathologist with NPAAC Module 1 and 3A Certification, MBBS, PhD, FRCPA, FFRCPA

Associate Professor Min Ru Qiu MBBS, PhD, FRCPA, FFRCPA, is an anatomical pathology trained molecular pathologist at SydPath, subspecialising in haematopathology and pulmonary pathology. A/Prof Qiu has been the lead molecular pathologist for many years and has extensive expertise in fluorescence in situ hybridization (FISH) assays, NGS and polymerase chain reaction (PCR)-based testing related to solid tumours and lymphoid malignancy. She was one of the first anatomical pathologists in Australia to receive the Certificate for Genetic/Genomic Testing from the RCPA in 2019.

She is actively involved with many external QAP programs. She is the convenor of the Oncology subcommittee of the RCPA QAP Molecular and Genome Advisory committee and has been an assessor of lung molecular modules of EMQN and GenQA. A/Prof Qiu is also a NATA technique assessor for molecular and genomic pathology. A/Prof Qiu has many collaborative projects within the campus research groups including the MoST study group, St Vincent's Centre for Applied Medical Research and Victor Chang Cardiac Research Institute. She has approximately 50 peer reviewed publications.

Associate Professor Tao Yang

Senior Molecular Anatomical Pathologist with NPAAC Module 3A, certification MBBS, PhD, FRCPA

Associate Professor Tao Yang MBBS, MSc, PhD, FRCPA, is a senior staff specialist in Anatomical Pathology and Molecular Pathology at SydPath. He is the co-lead molecular pathologist at SydPath and prior to his appointment, he was the Head of Molecular Pathology at Liverpool Hospital.

He was among the first pathologists in Australia to receive the Certification for Genetic/Genomic Testing from the Royal College of Pathologists of Australasia in 2019. A/Prof Yang collaborates with many clinicians and researchers and is recipient of both NHMRC and Cancer Council of New South Wales grants with research focuses on molecular mechanisms of cancers and their precursors.

Dr Stephen Wong

Molecular Anatomical Pathologist with NPAAC Module 1 Certification MBBS (Hons), FRCPA

Dr Stephen Wong BPharm (Hons) MBBS (Hons) FRCPA, is a staff specialist in Anatomical Pathology and Molecular Pathology at SydPath. He received the Certification for Genetic/Genomic Testing from the Royal College of Pathologists of Australasia in 2021. Dr. Wong possesses a wide-ranging passion for molecular pathology and is highly proficient in solid tumor fluorescence in situ hybridization (FISH) assays and next-generation sequencing (NGS). Dr. Wong is recognized as a national speaker and has delivered presentations at International Academy of Pathology conferences. Additionally, Dr. Wong has conducted educational webinars most notably for the RCPA QAP, focusing on the comparison between single gene testing and panel testing in non-small cell lung cancers.

Professor David Ma

Senior Molecular Haematopathologist with NPAAC Modules 2,3,4 Certification

Professor David Ma graduated in Medicine from the University of New South Wales. Following the completion of haematopathology training, he carried out several laboratory research studies overseas including those funded by RCPA Research Foundations in the ensuring years and has remained in this pursuit of scientific based medicine to the present. He is a senior specialist in Haematology at St Vincent's Public and Private Hospitals.

He is a Conjoint Professor of Medicine at the Clinical School, Faculty of Medicine, UNSW Sydney and, a member of the executive board and subcommittees of several international and national organisations. Professor Ma is also a Program Head of the St Vincent's Center for Applied Medical Research and the Director of R&D of the Haematology Department.

His laboratory research focuses on clinical and diagnostic applications of molecular and cell biology especially on haematopoietic and malignant stem cells. An example is that Professor Ma was instrumental in establishing the RCPA QAP for CD34 cell enumeration in the 2020's (subscribed by over 60 laboratories world-wide) and is currently working with the RCPA QAP team to set-up the world-first viable CD34 cell QAP programme. He has over 190 peer-reviewed publications (H-index 44 and over 11,000 citations). He is certified for karyotyping, FISH, SNP array, MLPA and NGS testing from the Royal College of Pathologists of Australasia.

Dr Michael Krigstein

(Molecular Haematopathologist with NPAAC Modules 2,3,4 Certification)

Dr Michael Krigstein MBBS(Hons) FRACP FRCPA MCncrSc is a staff specialist in Haematology at Sydpath. He holds certification for karyotyping, FISH, SNP array, MLPA and NGS testing in haematological malignancies. He is incredibly passionate about using molecular testing to accurately diagnose and prognosticate haematological malignancies to optimise therapeutic decision making. He has several publications in molecular haematology. He is also well recognised as an educator in the field, providing lectures for the Royal College of Pathologists of Australasia and teaching haematology trainees across the country.

Our Pathologists are supported by the dedicated and diligent SydPath team of scientists, technicians & administrative staff.

Dr Damien Stark

Principal Hospital Scientist BSc, MSc, PhD, FASM, FACTM, FFSc (Research) RCPA.

Dr Damien Stark is a Principal Hospital Scientist and current laboratory Manager of the NGS facility. He was instrumental in setting up the NGS laboratory and has over 25 years' experience as a medical scientist.

Dr Stark has over 120 peer reviewed scientific articles published (h-index 46) in various areas of microbiology, with a strong emphasis on molecular diagnostics.

Dr Bin Wang

Senior Hospital Scientist/Clinical Scientist

Dr Bin Wang MBBS is the clinical scientist who is the scientist in charge in the Molecular Pathology Lab/Cancer Genetic of SydPath. He has over 28 years of experience in molecular genetics with over 100 publications in world renowned journals and is a well-recognized expert in the field.

Dr Wang has played a pivotal role in achieving NATA accreditation for the multiple somatic mutation NGS panels that are currently offered at SydPath as well as managing the lab daily to maintain the quality of NGS services. Dr Wang has ongoing collaborations with researchers and clinicians alike and is a proud recipient of three national urgent initiative grants by the NHMRC for development of new genetic technologies and assays for viral, bacterial and cancer testing.

CURRENT NGS SERVICE CAPABILITIES

50 Gene Assay: This is considered our smaller gene panel which targets 50 key genes including EGFR, ALK, BRAF, ROS1, RET, KRAS, MET, and ERBB2, among others (listed in the table below). There is an option for DNA testing or RNA testing alone or in combination. This is our most commonly used panel and is considered suitable for malignant tumour types including lung cancer (DNA and/or RNA), colorectal carcinoma, melanoma, thyroid carcinomas, pancreatic carcinomas, and all other solid tumours including sarcomas.

DNA Hotspots					CNVs		Inter-Genetic Fusions		Intgra-Genetic Fusions
AKT1	CHEK2	FGFR3	KIT	NTRK3	ALK	FGFR1	ALK	NTRK1	AR
AKT2	CTNNB1	FGFR4	KRAS	PDGFRA	AR	FGFR2	BRAF	NTRK2	EGFR
AKT3	EGFR	FLT3	MAP2K1	PIK3CA	CD274	FGFR3	ESR1	NTRK3	MET
ALK	ERBB2	GNA11	MAP2K2	PTEN	CDKN2A	KRAS	FGFR1	NUTM1	
AR	ERBB3	GNAQ	MET	RAF1	EGFR	MET	FGFR2	RET	
ARAF	ERBB4	GNAS	MTOR	RET	ERBB2	PIK3CA	FGFR3	ROS1	
BRAF	ESR1	HRAS	NRAS	ROS1	ERBB3	PTEN	MET	RSPO2	
CDK4	FGFR1	IDH1	NTRK1	SMO			NRG1	RSPO3	
CDKN2A	FGFR2	IDH2	NTRK2	TP53					

161 Gene Assay: This is a multi-biomarker NGS assay that covers 161 of the most relevant cancer genes. This panel covers both DNA and RNA alterations with 87 hot spot genes, 43 focal CNV gains, 48 full coding regions for deletions and 51 fusion drivers. This is currently our most comprehensive genomic profiling panel for malignant tumours.

Hotspot Genes				Full-Length Genes			Copy Number Genes		Gene Fusions (Inter- and Intragenic)		
AKT1	ESR1	KIT	PDGFRB	ARID1A	FBXW7	PTEN	AKT1	FGFR4	AKT2	FGFR2	NUTM1
AKT2	EZH2	KNSTRN	PIK3CB	ATM	MLH1	RAD50	AKT2	FLT3	ALK	FGFR3	PDGFRA
AKT3	FGFR1	KRAS	PIK3CA	ATR	MRE11	RAD51	AKT3	IGF1R	AR	FGR	PDGFRB
ALK	FGFR2	MAGOH	PPP2R1A	ATRX	MSH6	RAD51B	ALK	KIT	AXL	FLT3	PIK3CA
AR	FGFR3	MAP2K1	PTPN11	BAP1	MSH2	RAD51C	AXL	KRAS	BRCA1	JAK2	PRKACA
ARAF	FGFR4	MAP2K2	RAC1	BRCA1	NBN	RAD51D	AR	MDM2	BRCA2	KRAS	PRKACB
AXL	FLT3	MAP2K4	RAF1	BRCA2	NF1	RNF43	BRAF	MDM4	BRAF	MDM4	PTEN
BRAF	FOXL2	MAPK1	RET	CDK12	NF2	RB1	CCND1	MET	CDKN2A	MET	PPARG
BTK	GATA2	MAX	RHEB	CDKN1B	NOTCH1	SETD2	CCND2	MYC	EGFR	MYB	RAD51B
CBL	GNA11	MDM4	RHOA	CDKN2A	NOTCH2	SLX4	CCND3	MYCL	ERBB2	MYBL1	RAF1
CCND1	GNAQ	MED12	ROS1	CDKN2B	NOTCH3	SMARCA4	CCNE1	MYCN	ERBB4	NF1	RB1
CDK4	GNAS	MET	SF3B1	CHEK1	PALB2	SMARCB1	CDK2	NTRK1	ERG	NOTCH1	RELA
CDK6	H3F3A	MTOR	SMAD4	CREBBP	PIK3R1	STK11	CDK4	NTRK2	ESR1	NOTCH4	RET
CHEK2	HIST1H3B	MYC	SMO	FANCA	PMS2	TP53	CDK6	NTRK3	ETV1	NRG1	ROS1
CSF1R	HNF1A	MYCN	SPOP	FANCD2	POLE	TSC1	EGFR	PDGFRA	ETV4	NTRK1	RSPO2
CTNNB1	HRAS	MYD88	SRC	FANCI	PTCH1	TSC2	ERBB2	PDGFRB	ETV5	NTRK2	RSPO3
DDR2	IDH1	NFE2L2	STAT3				ESR1	PIK3CB	FGFR1	NTRK3	TERT
EGFR	IDH2	NRAS	TERT				FGF19	PIK3CA			
ERBB2	JAK1	NTRK1	TOP1				FGF3	PPARG			
ERBB3	JAK2	NTRK2	U2AF1				FGFR1	RICTOR			
ERBB4	JAK3	NTRK3	XPO1				FGFR2	TERT			
ERCC2	KDR	PDGFRA					FGFR3				

BRCA Assay: Our BRCA assay provides 100% exonic coverage, including flanking intronic sequences (average of 64 bases in either direction) for both BRCA1 and BRCA2 genes. This panel has been validated for somatic mutation testing in prostate carcinoma.

Myeloid Assay: The Myeloid DNA panel is comprised of 45 key genes covering the most relevant targets associated with myeloid malignancies, including acute myeloid leukaemia (AML), myelodysplastic syndrome (MDS), myeloproliferative neoplasms (MPN) and myelodysplastic/myeloproliferative (MDS/MPN) overlap disorders. The assay detects somatic variants down to a 3% variant allele frequency, which can be useful for:

- Establishing accurate diagnoses (including differentiating malignant from non-malignant conditions)
- Providing a guide to prognosis
- Identifying therapeutic targets and molecular markers for ongoing monitoring

Each patient receives a personalised report incorporating the latest molecular classifications and knowledge. From November 1st 2023 this test is MBS subsidised for all patients with suspected myeloid malignancies at diagnosis, disease progression or relapse.

NGS CAPABILITIES COMING SOON

Lymphoma Assay

Genes		
ARID1A	FAM46C (TENT5C)	PIM1
ATM	FAS	PLCG2
B2M	FBXW7	POT1
BCL2	FOXO1	PRDM1
BCL6	GNA13	PTEN
BIRC3	HRAS	RHOA
BRAF	ID3	RPS15
BTK	IDH2	SAMHD1
CARD11	IRF4	SF3B1
CCND1	KMT2D/MLL2	SGK1
CCND3	KRAS	SMARCA4
CD79B	MAP2K1	SOCS1
CDKN2A	MEF2B	STAT3
CREBBP	MYC	STAT5B
CXCR4	MYD88	STAT6
DIS3	NFKBIE	TET2
DNMT3A	NOTCH1	TNFAIP3
EP300	NOTCH2	TNFRSF14
ETV6	NRAS	TP53
EZH2	PAX5	XPO1

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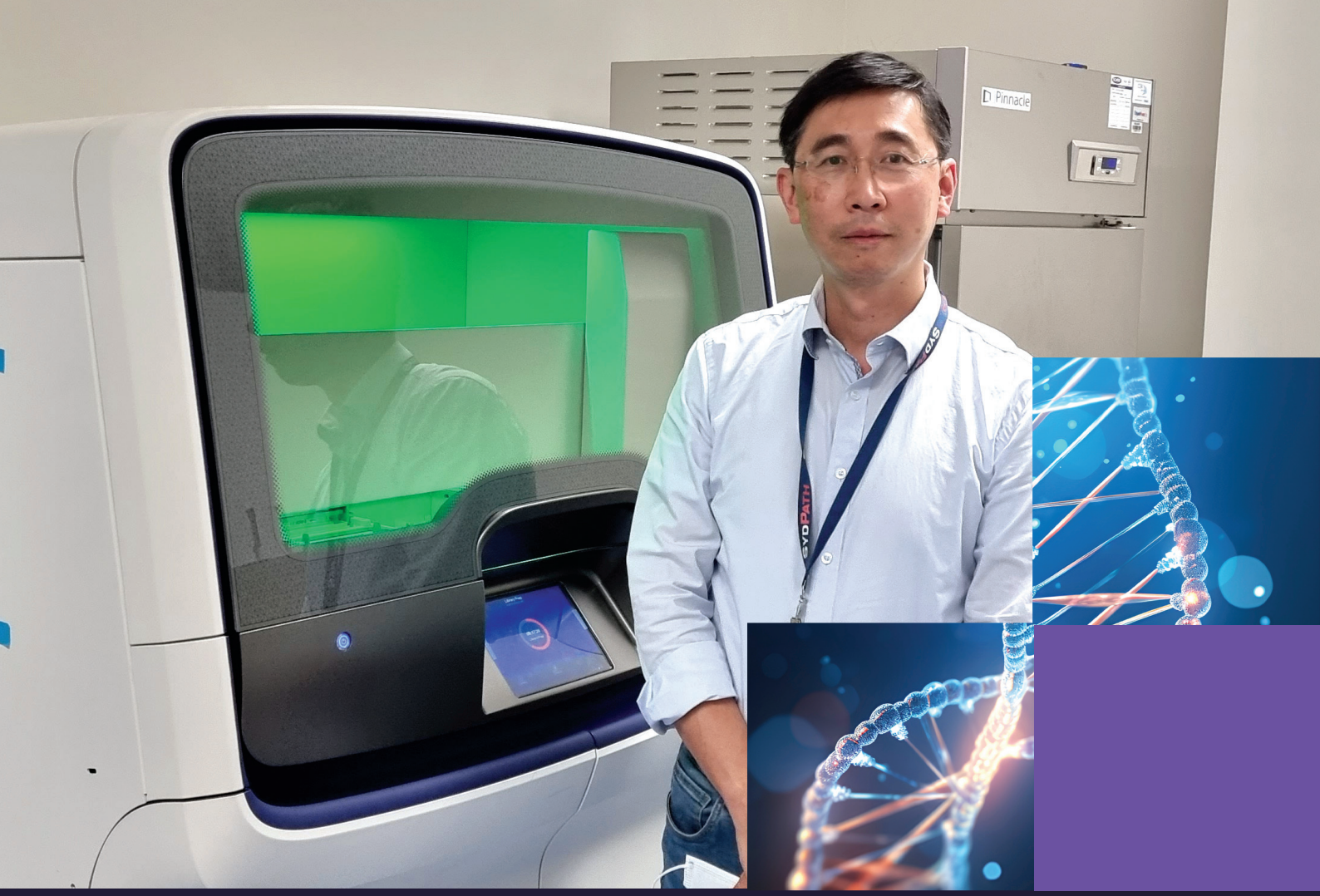
Our panel for lymphoid malignancies is currently undergoing validation and is expected to launch soon. It covers 60 genes that are crucial in B and T cell lymphomas. Once validated, this test will be fully MBS subsidised for all patients with suspected lymphoid malignancies at diagnosis, disease progression or relapse.

Cell-Free Assays for Liquid Biopsy Clinical Research: Cell-free nucleic acids assays are multibiomarker next-generation sequencing assays that enable reproducible detection of somatic mutations in plasma, down to a level of 0.1% in genes relevant to solid tumours. Optimised for liquid biopsy clinical research focused on primary driver and resistance mutations in cell-free nucleic acids. Currently this panel is in undergoing validation.

500+ Gene Assay: Comprehensive genomic profiling (CGP) is advancing precision oncology research through the analysis of multiple relevant biomarkers in a single NGS test.

- 500+ unique genes profiled for single gene and multi-gene biomarker insights
- Enables analysis of various single-gene variants, such as SNVs, indels, fusions, splice variants, and CNV, including both copy number gains and losses across 500+ genes
- Detection of complex biomarkers associated with immunotherapies such as TMB, MSI
- Support Homologous Recombination Deficiency (HRD) research by detecting mutations in 46 HRR genes, including BRCA LGRs, and assessment genomic scarring with the genomic instability metric (GIM)



ST VINCENT'S HOSPITAL SYDNEY

SydPath ST VINCENT'S PATHOLOGY

THE SYDPATH NGS PLATFORM IS SUPPORTED AND COMPLEMENTED BY OUR HIGH QUALITY ANATOMICAL PATHOLOGY AND HAEMATOLOGY MODALITIES INCLUDING HISTOLOGY, IMMUNOHISTOCHEMISTRY (INCLUDING PD-L1), CYTOGENETICS AND IN SITU HYBRIDISATION.

REQUEST FORMS

SydPath has a dedicated NGS testing request forms which contain many specific details including which panels are applicable to specific tumour types and contexts, which MBS items may be applicable and the costs for privately funded testing.

- **REQUEST FORMS - SYDPATH** WWW.SYDPATH.COM.AU/REQUEST-FORMS/
- **SYDPATH CANCER GENETICS SOMATIC MUTATION REQUEST FORM**
- **SYDPATH HAEMATOLOGY CANCER GENETICS REQUEST FORM**

CONTACT US

Please feel free to contact the SydPath NGS department or our Pathologists directly with any queries you might have about the somatic mutation testing and Next Generation Sequencing SydPath offers.

SydPath Pathologists
apoffice@svha.org.au

SydPath Next Generation Sequencing
sydpathcancer@svha.org.au

T **+61 2 8382 9100**

Level 6 Xavier Building,
St Vincent's Hospital, Victoria Street,
Darlinghurst Sydney NSW 2010 Australia
T +61 2 8382 9100
F +61 2 8382 2489
W sydpath.com.au

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