

Wellington Regional Genetics Laboratory (WRGL) is

one of New Zealand's major diagnostic laboratories, with a team of highly experienced staff and proven track record in delivery. At WRGL, we strive to support your clinical needs by providing timely cytogenetic services of the highest quality. Our Oncology team provides a first-class service with rapid turnaround times, well within NPACC professional guidelines. We aim to report our results as early as possible, and we will always prioritise urgent sample requests.

### **Turnaround times: 2017**

2017	Wellington mean (days)	Australasian mean (days)	NPAAC guidelines (days)
Bone Marrows	6.8	15	<b>18</b> urgent 5 days
FISH haematology	3.5	-	<b>18</b> urgent 5 days
FISH paraffin section	4.3	5.8	<b>18</b> urgent 5 days

# **User Satisfaction Survey**

Our latest user survey was conducted in April this year, with respondents rating our overall service:

- Outstanding (50% of respondents)
- Very satisfactory (50% of respondents)

As part of our continuous improvement programme, the Oncology team will be focusing on developing more inhouse molecular tests including Next Generation Sequencing (NGS), along with maintaining consistently low turnaround times.

### **New Tests**

We have recently validated several new tests, outlined below. The tests are free-of-direct charge for the Central Region DHBs covered by the Crown Funding Agreement.

### **ALK FISH Testing on FFPE samples**

WRGL now offers validated *ALK* FISH testing on FFPE samples. The anaplastic lymphoma kinase (*ALK*) gene has emerged as the second driver oncogene in lung cancer, for which highly effective novel therapies have been developed. FISH is the method of choice for detecting this rearrangement using the ALK break-apart probe for diagnosis of lung cancers with EML4-ALK rearrangement.

# Plasma Cell Sorting in Multiple Myeloma (MM)

WRGL routinely performs plasma cell sorting on MM cases. An abnormal karyotype is found in only 33% of patients with MM and interphase fluorescence *in situ* hybridisation (FISH) is the most useful test for studying chromosomal abnormalities.

Following plasma cell sorting, FISH studies show chromosomal abnormalities present in almost 90% of cases. For successful purification cell sorting is performed within 24hrs due to the fragility of plasma cells.

# **Tests in Development**

### MYD88 (L265P) Testing

Testing has been set up and validated in the molecular genetics laboratory at WRGL for the detection of the *MYD88* mutation using an allele specific PCR assay. The *MYD88* mutation is the hallmark change in patients with Waldenström macroglobulinemia and is also seen in splenic marginal zone lymphoma (SMZL) and in other B-cell chronic lymphoproliferative disorders.

Solid Tumours	Gene & Region			
Adenocarcinoma, Inflammatory	ALK (2p23)			
Myofibroblastic Tumour				
Adenocarcinoma	ROS1			
Solitary fibrous tumours	STAT6 (12q13)			
Alveolar soft part sarcoma X	TFE3 (Xp11.2)			
translocation renal cell carcinoma				
Dermatofibrosarcoma protuberta	ns COL1A1/PDGFB			
(DFSP)	((17;22)			
MM FISH Panel New & Existing				
CKS1B (1q21) Gain o	f 1q			
CDKN2C (1p32.3) Loss o	f 1p			
ATM/TP53 Loss o	f TP53			
D12Z3/D13S319/LAMP1 Deletions of 13q				
IGH rearrangement: Reflex				
t(4;14), t(11;14), t(14;16), t(14;20)	IGH/MAFB t(14;20)			

### **Recently Acquired FISH Probes**

For a comprehensive list of all FISH tests available please visit our website: www.wellingtongenetics.co.nz

### Please tell us what you want

Many FISH probes are commercially available and PCR or MLPA testing are also performed at WRGL. If there is any test you require, we are happy to discuss. Please contact Angela Brown (Section Head Oncology) angela.brown@ccdhb.org.nz

### Next Generation DNA Sequencing (NGS)

NGS has emerged as an accurate, cost-effective method to identify mutations across numerous genes known to be associated with response or resistance to specific targeted therapies. Genetic Laboratories are undergoing a technological revolution and the use of NGS will become mainstream within the next decade.

WRGL is involved in pilot schemes to develop this capability and we also have recruited a scientist with extensive NGS experience. We are currently validating this technique and haematological malignancy sequencing will follow.