

New Tests Now Available

Wellington Regional Genetics Laboratory
(WRGL) has validated testing for:

ALK FISH

JAK2 FISH

MYD88 mutation testing

The tests are free-of-direct charge for the Central Region DHBs covered by the Crown Funding Agreement.

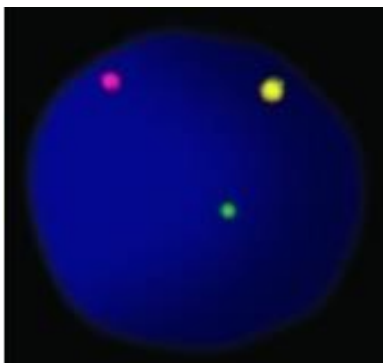
MYD88 (L265P) Mutation Testing

Testing has been validated in the molecular genetics section 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.

JAK2 (9p24.1) FISH probe

JAK2 rearrangements are found in Myeloid/lymphoid neoplasms with eosinophilia and have been included in the 2017 WHO as a provisional entity.

The *JAK2* gene is a protein tyrosine kinase involved in cytokine signalling. Chromosomal translocations involving *JAK2* can lead to the formation of chimeric onco-proteins in hematologic malignancies. Rearrangements involving 9p24.1 are typically aggressive and rare abnormalities seen in various hematologic diseases.



Vysis JAK2 breakapart probe

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.

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

Next Generation Sequencing (NGS) Update

WRGL is trialling the AmpliSeq for Illumina Cancer Hotspot Panel which is a targeted resequencing assay for mutations across the 50 genes with known associations to cancer types, including leukaemia, lung, colon, breast, ovarian, melanoma and prostate. Mutations in the hotspot regions of 50 oncogenes and tumour suppressor genes as identified in the COSMIC database (see table below).

**Table 2: AmpliSeq for Illumina Cancer Hotspot Panel v2
Genes**

<i>ABL1</i>	<i>EGFR</i>	<i>GNAS</i>	<i>KRAS</i>	<i>PTPN11</i>
<i>AKT1</i>	<i>ERBB2</i>	<i>GNAQ</i>	<i>MET</i>	<i>FB1</i>
<i>ALK</i>	<i>ERBB4</i>	<i>HNF1A</i>	<i>MLH1</i>	<i>RET</i>
<i>APC</i>	<i>EZH2</i>	<i>HRAS</i>	<i>MPL</i>	<i>SMAD4</i>
<i>ATM</i>	<i>FBXW7</i>	<i>IDH1</i>	<i>NOTCH1</i>	<i>SMARCB1</i>
<i>BRAF</i>	<i>FGFR1</i>	<i>JAK2</i>	<i>NPM1</i>	<i>SMO</i>
<i>CDH1</i>	<i>FGFR2</i>	<i>JAK3</i>	<i>NRAS</i>	<i>SRC</i>
<i>CDKN2A</i>	<i>FGFR3</i>	<i>IDH2</i>	<i>PDGFRA</i>	<i>STK11</i>
<i>CSF1R</i>	<i>FLT3</i>	<i>KDR</i>	<i>PIK3CA</i>	<i>TP53</i>
<i>CTNNB1</i>	<i>GNA11</i>	<i>KIT</i>	<i>PTEN</i>	<i>VHL</i>

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.

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