

Chronic lymphocytic leukemia (CLL) Service

Background

Chronic lymphocytic leukemia (CLL) is a hematological malignancy that is caused by excess B cell lymphocyte production in the bone marrow. It is the most common leukemia in the Western world and has its highest incidence in adults >80 years.

CLL patients are screened for variants in the TP53 and ATM genes as they provide prognosis and treatment indications. Between 5-10% of new CLL patients have deletions in the 17p chromosomal region or mutations in the TP53 gene (Schuch et al., 2018) These patients are not responsive to standard chemotherapy and instead they benefit from targeted therapy which is directed at the TP53 disruption. Patients with deletions in the ATM gene may require a more aggressive treatment regimen.

Test information

- **New and relapsed CLL cases and patients for treatment review and will receive a screen consisting of:**
 - Fluorescent in situ hybridisation (FISH) for the detection of mutations in the TP53 and ATM genes.
 - Next generation sequencing for the detection of mutations in the TP53 gene.

Referral Criteria

All requests should be made on an appropriate request form available at the AWMGS website www.medicalgenomicswales.co.uk.

<p>TAT (Calendar days)</p> <hr style="border: 1px solid black;"/> <p>FISH and NGS for CLL 21 cd</p>	<p>Contact Details</p> <p>All Wales Genomics Laboratory, Institute of Medical Genetics, University Hospital of Wales, Heath Park, Cardiff CF14 4XW</p> <p>Tel: 029 2074 2641 Fax: 029 2074 4043</p> <p>Haematology.Genetics.cav@wales.nhs.uk</p> <p>lab.genetics@wales.nhs.uk www.wales.nhs.uk/AWMGS/</p>
<p>Sample Requirements</p> <p>Peripheral blood in EDTA for FISH and NGS Please label samples with three identifiers and date of collection.</p> <p>All samples must be accompanied by a completed request form</p> <p>Consent for testing and sample storage is assumed when the request is received – it is the responsibility of the referring clinician to ensure that appropriate consent has been obtained</p>	

References

Schuh, A.H., Parry-Jones, N., Appleby, N., Bloor, A., Dearden, C.E., Fegan, C., Follows, G., Fox, C.P., Iyengar, S., Kennedy, B., McCarthy, H., Parry, H.M., Patten, P., Pettitt, A.R., Ringshausen, I., Walewska, R. and Hillmen, P. (2018). Guideline for the treatment of chronic lymphocytic leukaemia. British Journal of Haematology, 182(3), pp.344–359.