AWMGS
All Wales Medical Genomics Service

Waldenström Macroglobulinemia (WM) Service

Background

Waldenström Macroglobulinemia (WM) is a type of Non-Hodgkin's lymphoma also known as lymphoplasmacytic lymphoma. It is a lymphoproliferative disorder that gives an increased level of non-functional Immunoglobulin M in the blood, causing it to become thicker and slower moving. Clinical presentation of WM is extremely heterogeneous and can include; anaemia, lymphadenopathy and splenomegaly. Some WM patients however are entirely asymptomatic (smoldering WM).

The MYD88 L265P (OMIM: 602170.0004) mutation in exon 5 of the MYD88 gene is present in 91% of patients with WM (Treon et al. 2012), with the highest instance of WM occurring in older age groups aged 70+ (Wang et al. 2012). MYD88 c.794T>C L265P variant analysis is therefore used as a tool in the differential diagnosis of WM.

Test information

Digital Droplet PCR (ddPCR) is available for the detection of the MYD88 L265P variant. This method partitions samples into ~20,000 droplets, with PCR amplification being carried out on each of the droplets, creating a highly sensitive assay that can detect the L265P mutation at levels down to 1% in a background of genomic DNA.

MYD88 L265P variant date must be interpreted in combination of clinical and morphological features.

This service aims to offer a 14-calendar day turnaround time.

Referral Criteria

The assay is designed for use in patients with clinical features of Waldenström's Macroglobulinemia.
Referrals which do not meet this clinical criteria will not be processed.
This test is not suitable for minimal residual disease monitoring. DNA will be extracted and banked for all MRD requests.
All requests should be made on an appropriate request form available at the AWMGS website www.medicalgenomicswales.co.uk.

Sample Requirements

Bone marrow - send in sterile transport medium supplied by laboratory or in a lithium heparin blood tube.
FFPE - tissue scrolls
Blood - in EDTA

Please label samples with three identifiers and date of collection
All samples must be accompanied by a completed request form
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.

Links for further Information

OMIM: http://www.omim.org/
Genetic Test Registry: http://www.ncbi.nlm.nih.gov/gtr
Cancer Research UK: http://www.ncbi.nlm.nih.gov/gtr
All Wales Genomics Laboratory (AWGL):
Phone: 02920742641 Fax: 02920744043
Email: haematology.genetics.cav@wales.nhs.uk
Website: http://medicalgenomicswales.co.uk/

Contact Details

All Wales Genomics Laboratory,
Institute of Medical Genetics,
University Hospital of Wales,
Heath Park,
Cardiff CF14 4XW

Tel: 029 2074 2641
Fax: 029 2074 4043

Haematology.Genetics.cav@wales.nhs.uk
.
lab.genetics@wales.nhs.uk
www.wales.nhs.uk/AWMGS/

References

Treon, S.P., Xu, L., Yang, G., Zhou, Y., Liu, X., Cao, Y., Sheehy, P., Manning, R.J., Patterson, C.J., Tripsas, C., Arcaini, L., Pinkus, G.S., Rodig, S.J., Sohani, A.R., Harris, N.L., Laramie, J.M., Skifter, D.A., Lincoln, S.E. and Hunter, Z.R. (2012). MYD88 L265P Somatic Mutation in Waldenström's Macroglobulinemia. *New England Journal of Medicine*, [online] 367(9), pp.826–833.

Wang, H., Chen, Y., Li, F., Delasalle, K., Wang, J., Alexanian, R., ... & Wang, M. (2012). Temporal and geographic variations of Waldenstrom macroglobulinemia incidence: a large population-based study. *Cancer*, *118*(15), 3793-3800.