AWMGS
All Wales Medical Genomics Service

Droplet digital PCR for Systemic Mastocytosis
C-KIT p.(D816V) Variant

Background

Systemic Mastocytosis (SM) belongs to a group of myeloproliferative neoplasm characterised by the growth and accumulation of neoplastic mast cells. SM also involves at least one extracutaneous organ and may involve multiple hematopoietic cell lineages. The *KIT* D816V mutation is detectable in >80% of all patients with SM and is one of the minor criteria for a diagnosis of SM according to the 2016 updated World Health Organization classification of myeloproliferative neoplasms.

The All Wales Medical Genetics Laboratory (AWMGL) has validated the ddPCR protocol that detects the presence of the KIT D816V mutation. This method uses oil droplets to capture individual strands of DNA for amplification creating a highly sensitive assay that can detect the D816V mutation at levels of 0.1% in a background of wild-type genomic DNA. This is a diagnostic service for SM.

Referral Criteria

The assay is designed for use in patients with clinical features of Systemic Mastocytosis.

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.

The turn-around time is 10 working days.

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

Contact Details

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Sample Requirements

Bone marrow: Please send a minimum of 2ml in an EDTA tube.

Peripheral blood will be accepted from patients where BM sample is not available/appropriate, but may be more likely to give a false negative result.

Please label all samples with a minimum of 3 identifiers to include name and date of birth.