

Haemato-Oncology SNP Array Service

Background

Single Nucleotide Polymorphism (SNP) arrays are widely used in haemato-oncology to provide detailed information about the genetic changes that drive blood cancers. Unlike conventional karyotyping, SNP arrays offer high-resolution, genome-wide analysis, allowing detection of submicroscopic copy number changes and copy-neutral loss of heterozygosity that are clinically relevant for diagnosis and prognosis. SNP arrays support more accurate classification of disease, risk assessment and treatment planning, and can be equated to G-banding.

Test information

The following will receive SNP arrays where cytogenetic testing is requested:

- New or query **Acute Lymphoblastic Leukaemia (ALL)** cases
- New or query **Myelodysplastic Syndromes (MDS)** cases
- New **Myeloproliferative Neoplasms (MPN)** cases
- New **Chronic Myelomonocytic Leukaemia (CMML)** cases
- **Acute Myeloid Leukaemia (AML)** cases where conventional karyotyping has previously failed

Array Platform: Illumina Infinium® CytoSNP 850K v1.4 BeadChip Array.

Software: Illumina BlueFuse Multi v4.5.

Genome build: GRCh38.

Sensitivity:

- **10%-15% (clones/subclones lower than this proportion may not be detected).**
- Copy number changes below 5Mb or regions of copy neutral loss of heterozygosity below 10Mb are not reported unless affecting a disease-relevant gene/region†.

Limitations:

- **The array cannot detect balanced rearrangements or point mutations.**
- This test is unable to distinguish between germline and somatic findings.
- This test is not yet accredited to UKAS ISO 15189.

Sample requirements

- Bone marrow preferred (peripheral blood if BM unavailable).
- Please send 3ml in sterile transport medium or EDTA.
- Please label samples with three identifiers and date of collection.
- All samples must be accompanied by a completed request form.

PLEASE INCLUDE ALL RELEVANT CLINICAL DETAILS SUCH AS BLAST % AND MORPHOLOGY.

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.

Referral criteria

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

Turnaround times (calendar days)

ALL: 14CD

MDS, MPN, CMML: 21CD

Contact details

All Wales Medical Genomics Service, Wales Genomic Health Centre, Cardiff Edge Business Park, Longwood Drive, Whitchurch, Cardiff, CF14 7YU Haematology.genetics.cav@wales.nhs.uk

† Copy number changes below 5Mb will only be reported if they are detected within disease-relevant genes as listed below in accordance with referenced guidelines. Abnormalities <5Mb will otherwise not be interpreted or reported. Copy neutral loss of heterozygosity (CN-LOH) will only be reported if >10Mb and extending to the telomeres, to prevent incidental germline findings, except for CN-LOH of 17p/TP53. If there is clinical suspicion of germline disease association, please contact the laboratory to discuss further.

ALL	AML	MDS	MPN/CMML
<i>BTG1</i>	<i>EZH2</i>	<i>DNMT3A</i>	<i>MECOM</i>
<i>CDKN2A</i>	<i>MYD</i>	<i>MECOM</i>	<i>EZH2</i>
<i>CDKN2B</i>	<i>KMT2A</i>	<i>NPM1</i>	<i>KMT2A</i>
<i>EBF1</i>	<i>WT1</i>	<i>EZH2</i>	<i>TP53</i>
<i>ETV6</i>	<i>FLT3</i>	<i>CUX1</i>	<i>ASXL1</i>
<i>IKZF1</i>	<i>TP53</i>	<i>KMT2A</i>	<i>RUNX1</i>
<i>PAX5</i>	<i>ASXL1</i>	<i>ETV6</i>	
<i>RB1</i>	<i>RUNX1</i>	<i>TP53</i>	
PAR1 region	<i>ZRSR2</i>	<i>ASXL1</i>	
	<i>BCOR</i>	<i>RUNX1</i>	
	<i>STAG2</i>		

NHSE Haemato-oncology SNP array guidelines, 2023

IPSS-R [Greenberg et al. 2012, PMID: 22740453](#)

IPSS-M [Bernard et al. 2022, PMID: 38319256](#)

[Aly et al. 2019, PMID: 31320321](#)

[Walter et al. 2021](#)

[Gurney et al. 2023, PMID: 37144345](#)

[Walker et al. 2019, PMID: 31375516](#)

WHO 5th ed. [Khoury et al. 2022, PMID: 35732831](#)

ELN [Dohner et al. 2022, PMID: 35797463](#)

ICC [Weinberg et al. 2023, PMID: 36264379](#)

MIPSS70+ [Tefferi et al. 2018, PMID: 29708808](#)

CPSS [Elena et al. 2016, PMID: 27385790](#)