UK NEQAS
Leucocyte Immunophenotyping

Sheffield Teaching Hospitals NHS
NHS Foundation Trust

Pilot Myeloproliferative Neoplasms (MPN) Diagnostic Testing (Not Accredited)

Distribution – 232401 Participant -

Date Issued – 19 Jun 2023 Closing Date - 11 Aug 2023

Please note, this programme was previously titled Myeloproliferative Neoplasms Gene Panels (Pilot - Not accredited). It is designed for laboratories performing Myeloproliferative Neoplasms (MPN) testing using current algorithms[1] to diagnose and subtype the disease. Participants are expected to test samples according to their current testing pathways, and from April 2022 this programme has been limited (according to WHO/NCCN guidance for the testing of MPN patients) to the core clinically significant MPN variants: *JAK2* p.Val617Phe and clinically significant variants within *JAK2* exon 12, *CALR* exon 9 and *MPL* exon 10. Testing of all four regions is not mandatory; testing should be performed according to laboratory strategy, as well as test repertoire. Extended next generation sequencing panel data is no longer included in this programme and cannot be submitted. External quality assessment of such testing is now encompassed by the Myeloid Gene Panels (Pilot – Not Accredited) programme.

Trial Comments

This trial was issued to 105 participants; 100 participants (95.2%) returned results. Of the five participants that did not return results, one pre-notified us of their intended non-return.

Sample Comments

A single sample of genomic DNA, MPN DT 110, was issued by UK NEQAS LI. Participants were informed that the sample was from a 45-year-old male patient with a high haematocrit and possible diagnosis of polycythaemia vera (PV). They were asked to analyse this sample according to their 'in house' strategy for testing patients with a suspected myeloproliferative neoplasm, limited to the core MPN variants: *JAK2* p.(Val617Phe), and clinically significant variants within *JAK2* exon 12, *CALR* exon 9 and *MPL* exon 10, with exon numbering according to the MANE Select (v1.0)[2] reference transcripts: NM_004972.4(*JAK2*), NM_004343.4(*CALR*), and NM_005373.3(*MPL)*.

Sample MPN DT 110

Did you detect the clinically significant ***JAK2* p.(Val617Phe)** variant in sample MPN DT 110: No

Did you detect a clinically significant ***JAK2* exon 12** variant in sample MPN DT 110: Yes

Did you detect a clinically significant ***CALR* exon 9** variant in sample MPN DT 110: No

Did you detect a clinically significant ***MPL* exon 10** variant in sample MPN DT 110: No

Your Results

Gene/Region	Your DNA sequence variant	Your protein variant	Other details
JAK2 p.(Val617Phe)			
JAK2 exon 12	c.1614_1616delinsATT	p.(His538_Lys539delinsGlnLeu)	
CALR exon 9			
MPL exon 10			

All Participant Results

Gene region/marker	Participants detecting a variant/total number who tested the gene	Consensus DNA sequence variant*	Consensus protein variant*	Median variant allele burden (%)+
JAK2 p.(Val617Phe)	1/97	No variant detected	No variant detected	n/a
JAK2 exon 12	88/93	c.1614_1616delinsATT	p.(His538_Lys539delins GlnLeu)	33.0
CALR exon 9	0/88	No variant detected	No variant detected	n/a
MPL exon 10	0/86	No variant detected	No variant detected	n/a

*Results returned by participants (at both the DNA and protein level) may have been harmonised to the equivalent Human Genome Variation Society (HGVS) approved nomenclature (http://varnomen.hgvs.org/)[3,4] during the compilation of 'All Participant Results' tables. Nomenclature is based on the MANE Select (v1.0)[2] reference transcript and genome build GRCh38. Protein nomenclature includes parentheses as it represents a prediction from analysis at the DNA level.
+Descriptive statistics calculated for any variant with >2 quantification data points. Percentage values quoted have been subjected to rounding up/down to 1 decimal place.

Your Performance

Performance	Performance status for this sample	Running performance	
		Satisfactory	Critical
n/a	n/a	n/a	n/a

Please note: this programme is not currently performance monitored. We will work towards a performance monitoring system as the programme develops.

Detailed breakdown of *JAK2* exon 12 variant(s) detected

MANE Select transcript: NM_004972.4

DNA sequence change	n	Nomenclature guidance comments (as applicable)
c.1614_1616delinsATT	63	Compliant with current HGVS recommendations[3,4].
c.1614_1616dleinsATT	1	Consensus variant described mostly compliant with HGVS recommendations but including a typographical error.
c.1614_1616delCAAinsATT	12	Consensus variant described, mostly compliant with HGVS recommendations. Listing the deleted nucleotide sequence is not endorsed as this creates a longer description with redundant information.
c.[1614C>A(;)1615_1616delinsTT]	1	Consensus variant described, however sequence changes affecting two or more consecutive nucleotides should be described collectively as a single delins unless method provides information regarding phase. Square brackets should not be used when phase is uncertain[4].
c.1614C>A; c.1615A>T; c.1616A>T	2	Consensus variant described, however sequence changes affecting consecutive nucleotides should be described collectively as a single delins unless method provides information regarding phase.
c.1615_1616delinsTT	2	Out of consensus result.
c.1615_1616inv	3	Out of consensus result.
SNP	1	Out of consensus result. No HGVS based nomenclature provided.
Not provided	3	No details of the exon 12 variant detected were provided.

Colour coding reflects the level of compliance with current HGVS recommendations[3,4]: **green** = fully compliant, **amber** = generally compliant with some omission(s) and **red** = nomenclature error(s)/ fails to comply with the recommendations. **See Trial Comments section for further discussion**.

Protein sequence change	n	Nomenclature guidance comments (as applicable)
p.(His538_Lys539delinsGlnLeu)	52	Compliant with HGVS recommendations[3,4]. Parentheses appropriately reflect the analysis of DNA and the predicted status of the protein level description.
p.(H538_K539delinsQL)	1	Compliant with HGVS recommendations[3,4] however, use of three letter amino acid codes is preferred. Parentheses appropriately reflect the analysis of DNA and the predicted status of the protein level description.
p.His538_Lys539delinsGlnLeu	6	Parentheses to indicate predicted status of the protein level description are absent (or positioned inappropriately) and should be used when DNA is the input material[3,4]. Use of three letter amino acid codes is preferred, as applicable.
p.H538_K539delinsQL	3	
(p.His538_Lys539delinsGlnLeu)	1	
His538_Lys539delinsGlnLeu	2	Absent letter prefix at the protein level. Please note current HGVS recommendations[3,4], which state that a letter prefix is mandatory to indicate the type of reference sequence used. Parentheses to indicate predicted status of the protein level description are absent and should be used when DNA is the input material.
p.[His538Gln;Lys539Leu]	5	Nomenclature symbol/syntax error at the protein level. Consensus change described as two separate variants, with square brackets indicating that the variants are known to be *in cis*. Changes involving two or more consecutive amino acids should be described as deletion/insertion variants (delins). Variants should only be described individually when they are separated by one or more amino acids. Parentheses to indicate predicted status of the protein level description are absent and should be used when DNA is the input material[3,4].
p.(His538Gln)(;)(Lys539Leu)	1	Out-of-consensus protein description. Consensus change described as two separate variants, with (;) used to indicate that phase is uncertain.
p.His538Gln, p.Lys539Leu	1	Out-of-consensus protein description. Consensus change described as two separate variants with no attempt to describe phase. Parentheses to indicate predicted status of the protein level description are absent and should be used when DNA is the input material[3,4].
p.(His_Lys539delinsGlnLeu)	1	Nomenclature error at the protein level. Missing / misleading / inappropriate positional information or symbol.
p.HisLys538GlnLeu	2	
p.(H538_K539>QL)	1	
p.(His538_Lys539delinsGlnLau)	1	Nomenclature error at the protein level. Erroneous amino acid.
p.(His538_Lys539delinsGluLeu)	1	
p.(His538Gln);p.(Lys539Ter);p.Lys539Ile	1	Out-of-consensus submission
p.(Lys539Leu)	4	
(p.Lys539Leu)	1	
SNP	1	Out of consensus result. No HGVS based nomenclature provided.
Not provided	3	No details of the exon 12 variant detected were provided

Colour coding reflects the level of compliance with current HGVS recommendations[3,4]: **green** = fully compliant, **amber** = generally compliant with some omission(s) and **red** = nomenclature error(s)/ fails to comply with the recommendations. **See Trial Comments section for further discussion.**

Assay Details

Participants were asked to provided details of the methodologies used for each of their MPN assays. Responses are summarised in the following tables. Please note that some participants did not respond to all questions and some provided more than one response for some elements of their testing. In addition, at least one laboratory sent elements of their MPN testing to an external laboratory, in line with their local policy.

PCR Type	***JAK2* p.(Val617Phe)**	***JAK2* Exon 12**	***CALR* Exon 9**	***MPL* Exon 10**
PCR for NGS	42	55	45	48
Allele Specific PCR	23	2	7	8
Droplet Digital PCR	10	1	-	2
Multiplex PCR	1	4	2	3
Real-Time PCR	19	3	1	8
Single PCR	-	7	23	4
Sanger sequencing	-	13	5	7
Melting curve analysis	-	7	3	4
Cold PCR	-	1	-	-
Not given	2	1	2	2

Analysis Type	***JAK2* p.(Val617Phe)**	***JAK2* exon 12**	***CALR* exon 9**	***MPL* exon 10**
NGS (Illumina)	31	40	34	37
NGS (ThermoFisher Ion Torrent)	10	13	10	11
NGS (Other)	1	2	1	1
Agarose Gel Electrophoresis	8	-	-	1
Capillary Electrophoresis	6	9	27	5
Digital PCR (Bio-Rad)	10	1	-	3
High Resolution Melt	1	8	3	5
Real-Time PCR Fluorescent Detection	29	2	3	11
Sanger Sequencing	-	17	9	11
Not given	1	2	1	1

Protocol Type / Kit	*JAK2* p.(Val617Phe)	*JAK2* exon 12	*CALR* exon 9	*MPL* exon 10
Agilent SureSelect Custom QXT Panel	2	2	2	2
Archer DX VariantPlex Myeloid Panel	1	1	1	1
BioRad PrimePCR ddPCR Kit	8	-	-	-
Illumina AmpliSeq Panel	3	3	3	3
Illumina Custom Panel	1	1	1	1
Illumina TruSight Oncology 500	-	1	-	1
Illumina TruSight Myeloid Panel	1	2	2	2
In house Assay	41	51	53	40
MRC Holland SALSA MLPA Probemix P420	1	1	1	1
Qiagen QiaSeq Custom Panel	7	9	9	8
Qiagen QiaSeq Myeloid Neoplasms Panel	-	1	1	1
Qiagen/Ipsogen MutaQuant Kit	12	-	-	-
Qiagen/Ipsogen MutaScreen Kit CE	-	-	-	9
Qiagen/Ipsogen MutaSearch Kit	3	-	-	1
Qiagen/Ipsogen RGQ PCR Kit	-	-	3	1
Roche Kappa Capture	-	1	-	-
Roche Custom Panel	-	1	1	1
Rotor-Gene Q MDx	1	2	-	1
Sophia Genetics Myeloid Solution	3	3	3	3
Sophia Genetics Extended Myeloid Solution	1	2	1	1
ThermoFisher JAK2 V617F TaqMan SNP Assay	1	-	-	-
ThermoFisher Oncomine Myeloid Research Assay	7	7	5	7
ThermoFisher Oncomine Myeloid Gx v2 Panel	1	1	1	1
TibMolbiol LightMix JAK2 Exon 12 Assay	-	1	-	-
Other	1	1	-	-
Not given	2	2	1	1

Trial Comments

- Sample MPN DT 110 was comprised of genomic DNA from a 45-year-old male patient with a high haematocrit and possible diagnosis of PV.

***JAK2* Exon 12**

- Of the 100 participants returning results for this trial, 93 tested this sample for clinically significant variants in exon 12 of the *JAK2* gene. Two of the seven participants that did not perform *JAK2* exon 12 testing indicated that this testing is performed in an alternative laboratory. A further participant stated that their policy is to provide only *JAK2* p.(Val617Phe) testing for referrals that query a diagnosis of PV. The remaining four participants not providing a result for exon 12 of the *JAK2* gene did not comment further on their testing strategy.

- Eighty-eight participants (94.6% of those testing *JAK2* exon 12) reported a positive result, and overall, there was reasonable consensus in the variant detected, see below.
- Five participants (5.4%) reported an out-of-consensus negative result for *JAK2* exon 12 testing. Four laboratories used an in-house assay: either single PCR followed by capillary electrophoresis, multiplex PCR followed by capillary electrophoresis, real-time PCR with fluorescence detection or allele specific PCR (adapted from Furtado *et al*., 2013[5]) followed by capillary electrophoresis. Of interest, Furtado *et al.* acknowledge their assay may have a reduced ability to detect the consensus variant due to mismatch within the primer sequence. The remaining laboratory used the MRC Holland SALSA MLPA Probemix P420, which only detects two *JAK2* exon 12 variants and does not include the consensus variant observed in sample MPN DT 110.

***JAK2* p.(Val617Phe)**

- Ninety-seven participants tested this sample for the *JAK2* p.(Val617Phe) variant. Of the three participants who did not provide a *JAK2* p.(Val617Phe) result, one indicated that *JAK2* p.(Val617Phe) and exon 12 testing is performed at an alternative laboratory. The other two participants did not provide details of their testing strategy; however, one correctly identified the presence of a *JAK2* exon 12 variant in this sample, whilst the other provided an out-of-consensus negative result for their in-house multiplex PCR *JAK2* exon 12 assay.
- Of the 97 participants submitting a result for *JAK2* p.(Val617Phe) testing, 96 (99.0%) returned a negative result. The single participant returning an out-of-consensus positive result employed an in-house allele specific PCR assay (adapted from Klampfl *et al*., 2013[6]) followed by capillary electrophoresis, and reported a *JAK2* p.(Val617Phe) variant allele frequency (VAF) of 12.37%. This participant also provided an out-of-consensus negative result for their allele specific PCR *JAK2* exon 12 assay, raising the possibilities of a sample transposition event or an error at data entry.

***CALR* Exon 9**

- Eighty-eight participants tested this sample for clinically significant variants in exon 9 of the *CALR* gene. In line with expectation, all 88 (100%) returned a negative result.
- Twelve participants did not perform *CALR* exon 9 testing. Of these, three participants indicated that their laboratory does not standardly perform *CALR* exon 9 testing for patients referred with a possible diagnosis of PV, while a fourth participant stated that *CALR* exon 9, *JAK2* exon 12 and *MPL* exon 10 testing is performed at an alternative laboratory. The remaining eight centres did not provide further details of their testing strategies, however all eight reported a clinically significant variant in exon 12 of the *JAK2* gene. According to Tefferi and Pardanani (2014)[1], testing of *CALR* exon 9 is not required in PV patients following a positive result for *JAK2* p.(Val617Phe) or exon 12 testing.

***MPL* Exon 10**

- Eighty-six participants tested this sample for clinically significant variants in exon 10 of the *MPL* gene. In line with expectation, all 86 (100%) returned a negative result.
- Fourteen participants did not perform *MPL* exon 10 testing. Of these, two participants indicated that their laboratory does not standardly perform *MPL* exon 10 testing for patients referred with a possible diagnosis of PV, while two further participants stated that *MPL* exon 10 testing is performed at an alternative laboratory. The remaining ten centres did not provide further details of their testing strategies, however nine reported a clinically significant variant in *JAK2* exon 12, whilst one also did not test exon 12 of the *JAK2* gene. Again, according to Tefferi and Pardanani (2014)[1], testing of *MPL* exon 10 is not required in PV patients following a positive result for *JAK2* p.(Val617Phe) or exon 12 testing.

NM_004972.4 (*JAK2*): c.1614_1616delinsATT p.(His538_Lys539delinsGlnLeu)

- In line with expectation, 88/93 (94.6%) participants analysing exon 12 of the *JAK2* gene detected a clinically significant variant.
- Three participants indicated that a variant had been detected but did not describe the variant further; it is acknowledged that laboratories performing allele sizing or melt curve based assays to analyse *JAK2* exon 12 will lack sufficient sequence information from their results to provide HGVS based nomenclature. A fourth participant indicated that a SNP (single nucleotide polymorphism) had been detected but provided no further details for their out-of-consensus result. Please note that HGVS recommendations discourage the use of 'polymorphism'; 'variant' should be used in preference[3,4].

- For those 84 participants providing sequence information for this in-frame delins variant, application of the HGVS recommendations[3,4] for the description of sequence variants was variable.
- Sixty-four participants (76.2%) correctly described the cDNA change as c.1614_1616delinsATT.
- A further 12 participants (14.3%) reported the variant as c.1614_1616delCAAinsATT, which accurately describes the cDNA change, however listing the deleted nucleotide sequence is not endorsed as this creates a longer description with redundant information[3,4].
- One participant attempted to describe the delins variant as two separate events with uncertain phase: c.[1614C>A(;)1615_1616delinsTT]. Despite this format being advocated in the latest publication of the recommendations (Den Dunnen *et al*., 2016)[3], the current online version (20.05)[4] of the recommendations now states that use of '[ ]' is not correct in this scenario, instead c.1614C>A(;)1615_1616delinsTT should be used.
- Two participants described the delins variant by listing three single substitution variants: c.1614C>A; c.1615A>T; c.1616A>T.
- Five participants returned out-of-consensus HGVS-based results. Two centres incorrectly described the variant as a delins affecting only two of the three consensus nucleotides, c.1615_1616delinsTT, while three participants provided an alternative c.1615_1616inv description for the same cDNA change. Whilst c.1615_1616delinsTT and c.1615_1616inv both describe the substitution of AA with TT at c.1615_1616, please note that the latest publication of HGVS recommendations[4] prioritises the use of 'inv' over 'delins' when appropriate.
  Universal adoption of accurate prioritisation is important to help avoid variants being missed during literature searches and database queries.
  All five participants indicated that the variant results only in substitution of lysine with leucine at position 539, see below. NM_004972.4 (*JAK2*): c.1615_1616inv p.(Lys539Leu) is a commonly reported *JAK2* exon 12 variant[7,8] (COSMIC Genomic Mutation ID: COSV67575806; Legacy Identifier COSM24439)[9]. It is understood that, dependent upon the methodology used as well as primer positioning, a variant hotspot assay set up to detect c.1615_1616inv may give a positive result for sample MPN DT 110, without indication that the sample is positive for the longer variant c.1614_1616delinsATT. Two of these five participants used high resolution melting curve analysis and one used a multiplex PCR approach followed by capillary electrophoresis. It is less clear why the remaining two participants, both employing Sanger sequencing, may have returned this out-of-consensus result.

- There was further variation in the HGVS nomenclature submitted to describe the predicted protein change.
- Fifty-two participants (61.9%) provided the correct consensus protein description: p.(His538_Lys539delinsGlnLeu), and a further participant provided the consensus change but used single letter amino acid codes (3-letter codes are preferred[3,4]).
- An additional 10 participants (11.9%) provided the consensus description but failed to use parentheses (n=9) or positioned them incorrectly (n=1). Please note, where gDNA represents the assay input material, parentheses should be included in the nomenclature description to indicate the amino acid change is predicted from DNA level data.
- Two participants did not include 'p.' (current HGVS recommendations state that a letter prefix is mandatory to indicate the type of reference sequence used[3,4]), nor parentheses.
- Seven participants described the variant as two separate substitution events at the protein level.
  - Five Ion Torrent NGS users indicated that the variants were *in cis* (on the same allele) but failed to include parentheses to indicate the predicted nature of the amino acid changes: p.[His538Gln;Lys539Leu]. All five correctly described the variant as a 3-nucleotide delins at the DNA level.

  - One participant using Sanger sequencing indicated that phase is unknown: p.(His538Gln)(;)(Lys539Leu). This laboratory described the cDNA change as c.[1614C>A(;)1615_1616delinsTT]. As discussed above, the square brackets have been used inappropriately, but it is also unclear why the nucleotide changes at c.1615 and c.1616 were considered together, while the phase of the change at c.1614 was deemed uncertain.
  - Finally a further Sanger sequencing user merely listed two separate variants but did not include parentheses or attempt to describe phase: p.His538Gln, p.Lys539Leu. This participant also listed all nucleotide changes separately at the DNA level: c.1614C>A, c.1615A>T, c.1616A>T.
- As described above, five out-of-consensus participants indicated that the variant results only in substitution of the lysine residue at position 539 with leucine: p.(Lys539Leu).
- The remaining seven participants provided non-compliant nomenclature with a range of errors including incomplete positional numbering and using an erroneous symbol or amino acid code. One such submission was from a participant (using Sanger sequencing) who considered each nucleotide substitution independently and then provided conflicting predictions of the protein changes resulting from those substitutions: p.(His538Gln); p.(Lys539Ter); p.Lys539Ile.

- We recognise that different methodologies provide varying levels of information regarding the phase of sequence alterations, and that where phase ambiguity exists, application of HGVS recommendations to describe DNA and protein changes is particularly challenging. We are seeking clarification from the HGVS for scenarios where elements of a variant have previously been reported in isolation, however, as illustrated by Wakeling *et al*. (2020), failure to consider a multi-nucleotide variant correctly (in this case as a single delins event, rather than separate single nucleotide variants) can result in misannotation with the potential for false positive and false negative results. **NGS users should ensure that their analysis pipelines correctly annotate multi-nucleotide variants**[10].

- The *JAK2* c.1614_1616delinsATT p.(His538_Lys539delinsGlnLeu) variant identified in this trial has been previously reported in the literature in association with PV, albeit with some differing approaches to nomenclature[7,11,12] (COSMIC Genomic Mutation ID: COSV 67608841; Legacy Identifier COSM24438)[9]. Clinically significant variants in exon 12 of the *JAK2* gene are observed in ~3% of patients with PV[13,14] and detection of a clinically significant variant in exon 12 of the *JAK2* gene is included within the diagnostic criteria for PV[14].

Quantification of the *JAK2* exon 12 variant

- Fifty-eight participants provided a quantitative value for the *JAK2* exon 12 variant: variant/(variant+wild type)x100. Values (rounded to a single decimal place) ranged from 6.0% to 38.5% with a median value of 33.0% and interquartile range of 3.4%.
- NGS was the most common methodology, used by 54 participants (93.1%) with a median variant allele burden of 33.0% (range: 6.0% - 38.0%; IQR 3.1%).
- The remaining four participants used Sanger sequencing (n=2), droplet digital PCR (n=1) or did not provide details of their methodology (n=1).

Methodology

- Overall, the most commonly employed analysis methods were:
  - NGS (n=42, 43.3%), followed by Real-Time PCR with fluorescent detection (n=29, 29.9%) for *JAK2* p.(Val617Phe)
  - NGS (n=55, 58.5%), followed by Sanger sequencing (n=17, 18.1%) for *JAK2* exon 12 variants
  - NGS (n=45, 51.1%), followed by capillary electrophoresis (n=27, 30.7%) for *CALR* exon 9 variants
  - NGS (n=49, 57.0%), jointly followed by Sanger sequencing and Real-Time PCR with fluorescent detection (both n=11, 12.8%) for *MPL* exon 10 variants.
- Thus, a wide range of methods were used to detect variants in the core MPN associated genes, the majority of which have an adequate theoretical limit of detection (LoD) in the context of MPNs.

- Recent good practice guidelines advocate the ability to detect 1-3% VAF or lower for *JAK2* p.(Val617Phe) and 5% VAF for *JAK2* Exon 12, *CALR* exon 9 and *MPL* exon 10 variants[13]. Participants were asked to provide their LoD for each of their assays and the responses are summarised in the table below. This demonstrates that a subset of laboratories (highlighted red) are still using techniques with an inadequate LoD; transition to more sensitive techniques is therefore highly recommended for those centres.

Limit of Detection	*JAK2* p.(Val617Phe)	*JAK2* exon 12	*CALR* exon 9	*MPL* exon 10
<1%	35	1	3	2
1-3%	40	30	34	35
>3%/<5%	1	2	2	2
5%	11	39	32	28
>5%	1	14	6	9

Final comments

- We were aware that a minority of participants, most often those employing a single gene testing strategy, previously found the amount of DNA issued in this programme too small for their testing requirements. The amount issued in this trial was increased and appears to have been sufficient for the vast majority of laboratories. Whilst we are constrained by the size of the donated patient sample, we will endeavour to continue to issue a similar amount of DNA in future trials. Spare trial samples are generally available, and we encourage participants to request repeat samples when necessary (repeatsamples@uknegasli.co.uk).
- Please note that the number of trial issues for this programme has now increased from one to two per annum. This change took effect for the current 2023-2024 registration period; the second trial, MPN DT 232402, is scheduled for issue in January 2024.
- As panel testing of *JAK2*, *CALR* and *MPL* becomes more prevalent in the diagnosis of myeloproliferative neoplasms, UK NEQAS LI has begun the process of merging the current *JAK2* p.Val617Phe Mutation Status and MPN Diagnostic Testing Programmes to provide a more cost effective service whilst maintaining the rigour of the current programmes. It is intended that this process will be completed for the 2024-2025 registration period; further information will be provided in due course.
- We would like to thank laboratories for their continued participation in the pilot MPN Diagnostic Testing (Not Accredited) programme.

References

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Information with respect to compliance with standards BS EN ISO/IEC 17043:2010

4.8.2 a) The proficiency testing provider for this programme is:
UK NEQAS for Leucocyte Immunophenotyping
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4.8.2 b) The coordinators of UK NEQAS LI programmes are Mr Liam Whitby (Director) and Mr Stuart Scott (Centre Manager).

4.8.2 c) Person(s) authorizing this report:
Mr Liam Whitby (Director) or Mr Stuart Scott (Centre Manager) of UK NEQAS LI.

4.8.2 d) Pre issue testing of samples for this programme is subcontracted, although the final decision about sample suitability lies with the EQA provider; no other activities in relation to this EQA exercise were subcontracted. Where subcontracting occurs, it is placed with a competent subcontractor and the EQA provider is responsible for this work.

4.8.2 g) The UK NEQAS LI Confidentiality Policy can be found in the Quality Manual which is available by contacting the UK NEQAS LI office. Participant details, their results and their performance data remain confidential unless revealed to the relevant NQAAP when a UK participant is identified as having performance issues.

4.8.2 i) All EQA samples are prepared in accordance with strict Standard Operational Procedures by trained personnel proven to ensure homogeneity and stability. Where appropriate/possible EQA samples are tested prior to issue. Where the sample(s) issued is stabilised blood or platelets, pre and post stability testing will have proved sample suitability prior to issue.

4.8.2 l), n), o), r) & s) Please refer to the UK NEQAS LI website at www.ukneqasli.co.uk for detailed information on each programme including the scoring systems applied to assess performance (for BS EN ISO/IEC 17043:2010 accredited programmes only). Where a scoring system refers to the 'consensus result' this means the result reported by the majority of participants for that trial issue. Advice on the interpretation of statistical analyses and the criteria on which performance is measured is also given. Please note that where different methods/procedures are used by different groups of participants these may be displayed within your report, but the same scoring system is applied to all participants irrespective of method/procedure used.

4.8.2 m) We do not assign values against reference materials or calibrants.

4.8.2 q) Details of the programme designs as authorized by The Steering Committee and Specialist Advisory Group can be found on our website at www.ukneqasli.co.uk. The proposed trial issue schedule for each programme is also available.

4.8.2 t) If you would like to discuss the outcomes of this trial issue, please contact UK NEQAS LI using the contact details provided. Alternatively, if you are unhappy with your performance classification for this trial, please find the appeals procedure at www.ukneqasli.co.uk/contact-us/appeals-and-complaints/

4.8.4) The UK NEQAS LI Policy for the Use of Reports by Individuals and Organisations states that all EQA reports are subject to copyright, and, as such, permission must be sought from UK NEQAS LI for the use of any data and/or reports in any media prior to use. See associated policy on the UK NEQAS LI website: http://www.ukneqasli.co.uk/eqa-pt-programmes/new-participant-information/