UK NEQAS
Leucocyte Immunophenotyping

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Pilot MPN Gene Panels 202101(not accredited)

Issue date: 09 Sep 2020
Closing date: 30 Oct 2020

Results for MPN Gene Panels 202101 sample **MPN GP 106** can only be submitted via the following web entry pages.

In response to participant feedback we are currently working to develop a method of retaining laboratory, platform and assay information which is unlikely to change frequently between trial distributions. For example, protocol details, targeted genes (region of interest) and reference sequences employed. Therefore, reducing the need to re-submit the same information at results return for each trial distribution. PLEASE PROVIDE THE INFORMATION REQUESTED ON THE WEB DATA ENTRY PAGES TO FORM THE BASIS OF YOUR LABORATORY RECORD. THANK YOU.

Please do not start to complete this survey until you have all of your data ready to enter.

Please report any (potentially) clinically significant intragenic and/or regulatory element changes such as point mutations, small insertion, deletion and duplications events. There is also the opportunity to report variants of unknown clinical significance. However, please DO NOT report any variant(s) considered to be benign/likely benign. The reporting of larger changes affecting genome architecture or copy number changes (>50kb) is not required. We acknowledge best practice in somatic variant interpretation is an evolving topic. However, for further details regarding the classification terminology currently utilised for this trial please see Li MM et al Standards and Guidelines for the Interpretation and Reporting of Sequence Variants in Cancer. J Mol Diagn. 19(1):4-23 (2017). Please only submit results applicable to the scope of this EQA programme.

We strongly encourage continued completion of the fields related to region of interest (ROI), achieved coverage and reference sequence details. Collecting this information is important; it helps us to investigate anomalous results (potential false negatives, alternative nomenclature etc). Any numerical fields must be completed using only decimal points to separate numbers, and not commas e.g. enter 6.3 not 6,3. Please do not enter symbols e.g. % into numerical fields.

Repeat samples are available for all programmes. In the event that your local quality control (QC) criteria are not met please contact UK NEQAS LI as soon as possible. Please do not submit results based on a suboptimal extraction. Requests for repeat samples should be made by emailing admin@ukneqasli.co.uk. Should this not be possible please telephone our Administration team on the number provided below.

Contact details:

UK NEQAS LI, Pegasus House, 4th Floor, 463A Glossop Road, Sheffield, S10 2QD, UK.
Tel: +44 (0) 114 267 3600, Fax: +44 (0) 114 267 3601.
e-mail: admin@ukneqasli.co.uk

website: http://www.ukneqasli.co.uk/

Throughout the survey * denotes a mandatory field

Participant ID *

What best describes your testing strategy *

- ☐ Sequential single gene testing
- ☐ Parallel single gene testing
- ☐ Targeted Next Generation Sequencing Panel Testing

NGS platform and assay information

Which Next Generation Sequencing (NGS) platform was used to analyse the samples for this trial? *

- ☐ Life Tech Ion PGM
- ☐ Life Tech Ion Proton
- ☐ Life Tech Ion S5
- ☐ Life Tech Ion S5 XL
- ☐ Roche FLX/G20
- ☐ Roche Junior
- ☐ Illumina GA/GAII
- ☐ Illumina HiSeq
- ☐ Illumina MiSeq
- ☐ Illumina MiniSeq
- ☐ Illumina NextSeq
- ☐ Illumina Novaseq
- ☐ Applied Biosystems/Life Tech SOLiD
- ☐ Pacific Biosciences PacBio RS
- ☐ Helicos Biosciences HeliScope
- ☐ Azco Biotech MAX-Seq Genome Sequencer

If applicable, which approach to targeted NGS best describes your gene panel? *

- ☐ Agilent Haloplex HS Panel
- ☐ Agilent ClearSeq AML HS Panel
- ☐ Agilent SureSelect Custom QXT Panel
- ☐ BIOO NEXTflex Custom
- ☐ Fluidigm Access Array

- [ ] Illumina TruSeq Amplicon - Cancer Panel
- [ ] Illumina TruSeq Custom Panel
- [ ] Illumina TruSight Myeloid Sequencing Panel
- [ ] In house (amplicon based)
- [ ] In house (capture based)
- [ ] Ion AmpliSeq Cancer Hotspot Panel
- [ ] Myeloid Solution, Sophia Genetics
- [ ] OGT SureSeq Myeloid panel
- [ ] Nextera Illumina TruSight Myeloid Sequencing Panel
- [ ] Oncomine Myeloid Research Panel
- [ ] Qiagen GeneRead DNAseq Targeted Panels Human Myeloid Neoplasms Panel
- [ ] Qiagen QiaSeq Custom Panel
- [ ] Raindance Thunderbolt Cancer Panel
- [ ] Raindance Thunderbolt Myeloid Panel
- [ ] _____

IMPORTANT: THE INFORMATION YOU PROVIDE ON THIS PAGE WILL FORM THE BASIS OF YOUR LABORATORY'S RECORD FOR THIS PROGRAMME AND WILL REDUCE THE REQUIREMENT TO RE-SUBMIT THE SAME DETAILS FOR EACH TRIAL DISTRIBUTION. There will be an opportunity to review the information held in your record at each MPN Gene Panels trial distribution and the chance to request changes to ensure the record accurately reflects your laboratory and its current practice in relation to this programme.

Which annotation database resources do you routinely use during your variant interpretation process? (select all that apply)

- [ ] COSMIC (Catalogue Of Somatic Mutations In Cancer)
- [ ] dbSNP (Short Genetic Variations, NCBI)
- [ ] ClinVar (NCBI)
- [ ] My Cancer Genome (Vanderbilt-Ingram Cancer Center)
- [ ] HGMD (The Human Gene Mutation Database)
- [ ] OMIM (NCBI)
- [ ] The Cancer Genome Atlas (TCGA)
- [ ] The Genome Aggregation Database (gnomAD)
- [ ] Varsome (Aggregation tool)
- [ ] WHO International Agency for Research on Cancer (IARC) TP53 Database
- [ ] _____

Genome Assembly e.g. GRCh37p13 *

- [ ] GRCh37/hg19
- [ ] GRCh38
- [ ]

What is your minimum Variant Allele Frequency (VAF) for reporting identification of a variant? Please provide as a percentage (%) value. *

e.g. 5 (do not include the % symbol)

Please provide any further relevant details of your testing strategy

Region of interest details

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IMPORTANT: We strongly encourage continued completion of the table below. Collecting this information is important; it helps us to investigate anomalous results. Moving forward we hope to retain a bank of information to reduce the need to return this information every trial.

Please provide details of the genes/regions sequenced for this trial:

	Gene	Target Region (e.g whole gene, exons 1-9)	Reference Sequence (e.g. NM_XXXXXX.X)	Comments
Gene 1				
Gene 2				
Gene 3				
Gene 4				

Gene 5				
Gene 6				
Gene 7				
Gene 8				
Gene 9				
Gene 10				
Gene 11				
Gene 12				
Gene 13				
Gene 14				
Gene 15				
Gene 16				
Gene 17				
Gene 18				
Gene 19				
Gene 20				

Gene 21				
Gene 22				
Gene 23				
Gene 24				
Gene 25				
Gene 26				
Gene 27				
Gene 28				
Gene 29				
Gene 30				
Gene 31				
Gene 32				
Gene 33				
Gene 34				
Gene 35				
Gene 36				

Gene 37

Gene 38

Gene 39

Gene 40

Gene 41

Gene 42

Gene 43

Gene 44

Gene 45

Gene 46

Gene 47

Gene 48

Gene 49

Gene 50

Gene 51

Gene 52

Gene 53				
Gene 54				
Gene 55				
Gene 56				
Gene 57				
Gene 58				
Gene 59				
Gene 60				

Quality Control (QC) for samples MPN GP 106

With regard to sample MPN GP 106, please indicate if coverage of the previously stated target region of interest (ROI)* in a given gene was NOT achieved in line with your local QC. If this table is left blank it will be assumed full coverage was achieved across all gene regions

	Gene	ROI coverage NOT achieved	Exon(s) involved	Reference Sequence (e.g. NM_XXXXXX.X)	Comments
Gene 1					
Gene 2					
Gene 3					
Gene 4					

Gene 5					
Gene 6					
Gene 7					
Gene 8					
Gene 9					
Gene 10					
Gene 11					
Gene 12					
Gene 13					
Gene 14					
Gene 15					
Gene 16					
Gene 17					
Gene 18					
Gene 19					
Gene 20					

Gene 21					
Gene 22					
Gene 23					
Gene 24					
Gene 25					
Gene 26					
Gene 27					
Gene 28					
Gene 29					
Gene 30					
Gene 31					
Gene 32					
Gene 33					
Gene 34					
Gene 35					
Gene 36					

Gene 37					
Gene 38					
Gene 39					
Gene 40					
Gene 41					
Gene 42					
Gene 43					
Gene 44					
Gene 45					
Gene 46					
Gene 47					
Gene 48					
Gene 49					
Gene 50					
Gene 51					
Gene 52					

Gene 53					
Gene 54					
Gene 55					
Gene 56					
Gene 57					
Gene 58					
Gene 59					
Gene 60					

Single Gene Testing - Assay information

If you perform single gene testing please select all markers that you routinely test in MPN cases: *

- ☐ JAK2 p.Val617Phe
- ☐ JAK2 Exon 12
- ☐ CALR Exon 9
- ☐ MPL Exon 10
- ☐ ASXL1
- ☐

For labs that perform single gene testing please provide method details for the genes that you analysed. If you test genes other than JAK2, CALR and MPL, please use the 'Other' boxes and prefix the method information with the gene/variant name.

PCR Type	Analysis Type	Protocol Type

JAK2 p.Val617Phe			
JAK2 Exon 12			
CALR Exon 9			
MPL Exon 10			
ASXL1			
Other 1			
Other 2			
Other 3			

Please provide any further relevant details of your testing strategy:

Did you detect a clinically significant variant in sample MPN 106? *

- ○ Yes
- ○ No

Please provide details of any clinically significant variants you identified in Samples MPN GP 106. Please do not report any variants that are benign or likely benign.

Gene	DNA sequence change (cDNA description) e.g. c.944C>T. or description of the variant as your technology allows e.g. 30bp Insertion in exon 12	Resulting amino acid change (protein description), if applicable	Variant Allele Frequency Percentage / Mutation Load, if applicable	Annotation references (COSMIC, ClinVar, dbSNP etc), if applicable	Clinical significance of variant, if applicable	Read depth at variant position

Variant 1							
Variant 2							
Variant 3							
Variant 4							
Variant 5							
Variant 6							
Variant 7							
Variant 8							
Variant 9							
Variant 10							
Variant 11							
Variant 12							
Variant 13							
Variant 14							
Variant 15							
Variant 16							

Variant 17						
Variant 18						
Variant 19						
Variant 20						

If you wish to receive a confirmation email that your results have been received by UKNEQAS LI insert your e-mail address here

example@example.com

Please confirm your participant ID. *

Thank you, you have now completed your results return for MPN GP 202101

PLEASE NOTE: This is your last opportunity to review your data before submitting it. You will not be able to return to this survey after you have selected the Submit button below. If you wish to review the results you have entered please make use of the Back button to review the information on previous pages.

Contact details
UK NEQAS LI, Pegasus House, 4th Floor, 463A Glossop Road, Sheffield, S10 2QD, UK.
Tel: +44 (0) 114 267 3600, Fax: +44 (0) 114 267 3601.
e-mail: admin@ukneqasli.co.uk
website: http://www.ukneqasli.co.uk/

Throughout the survey * denotes a mandatory field

Are you satisfied with the service provided by UK NEQAS LI?

Yes

Any further comments