PGDx elio™ tissue complete
IVD Report

PGDx Personal Genome Diagnostics ™

Sample Name: VELIO0214T_TMB2_X8

Tumor Type:	Other	**Run Quality Result:**	PASS	**Assay Name:**	PGDx elio tissue complete (03)
Sample Type:	Specimen	**Sample Quality Result:**	PASS	**Assay Version:**	1.0.0.11
Details:	(blank)	**Control Quality Result:**	PASS	**Platform Version:**	v1.3.2.3-4
Report Date:	2020-05-06	**Overall Case Quality Result:**	PASS	**Flow Cell ID:**	HLCKLBGX9

Please refer to the Complete Case Record (CCR) for details on quality metrics

Summary of Results

Variant Category	Observation(s)
Variants with Evidence of Clinical Significance	1
Variants with Potential Clinical Significance	41

Variants with Evidence of Clinical Significance[§]

Genomic Signatures		
Signature	**Status**	**Indications with Supporting Evidence**
Microsatellite Analysis	MSI-H	Solid Tumor

Sequence Mutation Analysis				
Gene	**Alteration**	**Consequence**	**MAF (%)**	**Indications with Supporting Evidence**
BRAF	V600E	Missense	18.8	CNS, Colon, GIST, Melanoma, NSCLC, Rectal, Thyroid

Amplification Analysis		
Gene	**Alteration**	**Indications with Supporting Evidence**
No Amplifications with Evidence of Clinical Significance were detected.		

Translocation Analysis			
Gene	**Alteration**	**Partner**	**Indications with Supporting Evidence**
No Translocations with Evidence of Clinical Significance were detected.			

These results are intended to provide pan cancer tumor profiling information for use by qualified healthcare professionals in accordance with professional guidelines and are not conclusive or prescriptive for labeled use of any specific therapeutic product. Qualified healthcare professionals should confirm the clinical diagnosis and all available information, including whether the sample is from a primary or a metastatic site.

[§]Variants with evidence of clinical significance appear on the first page based on the selected tumor type. Variants with evidence of clinical significance in alternative tumor types may appear on subsequent pages.

Rx only

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Variants with Potential Clinical Significance

Genomic Signatures	
Signature	**Score**
TMB Muts/Mb	44.6

Sequence Mutation Analysis			
Gene	**Alteration**	**Consequence**	**MAF (%)**
ASXL2	N26Kfs*7	Frameshift	8.6
BCL6	P191L	Missense	21.3
BLM	N23del	In-frame Deletion	27.7
BMPR1A	E256K	Missense	18.6
CDH1	L582del	In-frame Deletion	8.1
CDK12	N474Kfs*12	Frameshift	22.6
CHD4	V1368Gfs*6	Frameshift	8.0
CREBBP	L1346Cfs*30	Frameshift	23.6
DAXX	A226E	Missense	21.1
ERCC4	R864H	Missense	22.0
ERCC5	K917Nfs*65	Frameshift	28.5
ETV4	D72Vfs*14	Frameshift	9.9
FANCE	P197S	Missense	21.7
FBXW7	A626P	Missense	21.5
FH	K477dup	In-frame Insertion	45.0
GLI1	n/a	Splice Site Acceptor	21.8
H3F3B	E95Tfs*?	Frameshift	18.6
HNF1A	P291Qfs*51	Frameshift	24.2
INPP4B	G529W	Missense	23.1
KDM5C	R328Q	Missense	46.7
KMT2C	R2167Q	Missense	22.9
KMT2C	F2334V	Missense	20.9
MSH6	D649G	Missense	45.4
NSD1	A2127V	Missense	20.2
PDGFRB	V582M	Missense	24.0
PIK3CG	V168M	Missense	23.1
PIK3R1	K16Rfs*15	Frameshift	21.7
RANBP2	Q2754R	Missense	23.9
RASA1	L81V	Missense	54.5
RNF43	G659Vfs*41	Frameshift	53.6
ROS1	F518Pfs*8	Frameshift	49.4
RPS6KA4	L228F	Missense	60.7
SMARCA4	R801C	Missense	25.1
SOX2	A27T	Missense	20.7

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Sequence Mutation Analysis (Continued)

Gene	Alteration	Consequence	MAF (%)
SPTA1	D705Y	Missense	20.4
SUFU	G218C	Missense	16.7
TLR9	V233I	Missense	18.2
TP53	P191del	In-frame Deletion	23.2
TP53	K382Nfs*?	Frameshift	24.0
TYRO3	A217T	Missense	22.0
WT1	G148S	Missense	25.8

Amplification Analysis

Gene	Alteration
No Amplifications with Potential Clinical Significance were detected.	

Translocation Analysis

Gene	Alteration	Partner
No Translocations with Potential Clinical Significance were detected.		

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Results Definition

Variants with Evidence of Clinical Significance: Aligns to AMP/ASCO/CAP NGS reporting Guidelines for Tier IA.

Variants with Potential Clinical Significance: Aligns to AMP/ASCO/CAP NGS reporting Guidelines for Tier IB-IV evidence.

- AMP/ASCO/CAP NGS Reporting Guidelines: Li,Marilyn M.et al (2017). Standards and Guidelines for the Interpretation and Reporting of Sequence Variants in Cancer. The Journal of Molecular Diagnostics, Volume 19, Issue 1, 4-23.

Genomic Signatures

- TMB Score
  - Muts/Mb: the mutations per megabase sequenced within the PGDx elio tissue complete targeted region of interest.
- Microsatellite Analysis
  - MSI-H: Microsatellite Instability High.
  - MSS: Microsatellite Stable (MSS).
  - Indeterminate - microsatellite status: Tract unstable frequency $\leq$ 0.1 and mutation signature score $\geq$ 20 OR Tract unstable frequency $\geq$ 0.25 and mutation signature score $\leq$-10

Sequence Mutation Analysis

- MAF: Mutant Allele Fraction
- M1?: Start codon loss, translation of this codon will likely not occur making an amino acid change not applicable.

Amplification Analysis

- Amplification: an increase in copy number was detected.
- Indeterminate - amplification: tumor purity could not be estimated to support the detection of copy number alterations of this gene.

Translocation Analysis

- Gene Fusion: a translocation event was detected where gene coding strand continuity is maintained (this may result in transcription of the coding strand of one gene into the coding strand of another gene).
- Rearrangement: a translocation event was detected where gene coding strand continuity is not maintained (this may result in transcription of the coding strand of one gene into the template strand of another gene).

Coverage

- Indeterminate - coverage: Coverage of this region is not sufficient to determine results.

Sample Reporting

- No Result: 'No Result' may be reported if the sample, control, or run have not met quality standards resulting in the inability to report results (please see Complete Case Record (CCR) for details on data quality metrics).

Indication

- CNS: Central Nervous System
- GIST: Gastrointestinal Stromal Tumor
- NSCLC: Non-Small Cell Lung Cancer

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Disclaimer and Limitations of Approach

For mutations listed in the Variants with Potential Clinical Significance section of the PGDx elio tissue complete report, the clinical significance has not been demonstrated with this test.

For mutations listed in the Variants with Evidence of Clinical Significance section of the PGDx elio tissue complete report, please refer to the Report Interpretation Guide for information concerning conclusions about the level of clinical evidence.

Next generation sequencing approaches may provide incorrect sequence or mutational data due to insufficient coverage in specific regions of the genome, inability to distinguish highly related human sequences, and sequencing errors and the analysis of sequence specific alterations can also be hampered by limitations related to the tumor DNA. A negative result does not rule out the presence of a mutation below the limits of detection of the assay.

The information contained in this report does not constitute medical advice. These findings are not intended for selection of therapy. While this report is believed to be accurate and complete as of the date issued, Personal Genome Diagnostics Inc. is not responsible for updating the report to incorporate newly obtained information or new interpretations of the assay results contained in the report.