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Central Zone

Pathology and Laboratory Medicine Memorandum

To: NS, PEI, NB, and NFLD Laboratories, Central Zone Physicians

From: Dr. Wenda Greer, Director of Molecular Diagnostics
Dr. Weei Huang, Anatomical Pathology

Date: November 29, 2016

Subject: Next Generation Sequencing (NGS) mutation panels

As of December 12, 2016 the Molecular Diagnostics Laboratory will be offering two NGS mutation panels:

- The Illumina TruSight Tumor 15 panel
- The Illumina Trusight Myeloid panel

Each specimen will be sequenced with the entire appropriate NGS panel.
Our analysis pipeline will assess and report only the mutation status of genes in the panel that are relevant to the diagnosis in question as described below.
Information is available upon request concerning mutations on the non-reported genes.

Genes sequenced/analyzed:

1. Genes sequenced by the **Illumina TruSight Tumor 15 panel** (if only part of a gene is sequenced, the exons are indicated in brackets):

   AKT1 (3), BRAF (15), EGFR (12, 18-21), ERBB2 (17-21, 24, 26), FOXL2 (1), GNA11 (5), GNAQ (5), KIT (8-11, 13, 14, 17, 18), KRAS (2-4), MET (16, 18, 20), NRAS (2-4), PDGFRA (12, 14, 18), PIK3CA (9, 20), RET (16), TP53 (1-11)

   Genes analyzed for:

   Lung Cancer
   EGFR (12, 18-21), KRAS (2-4), BRAF (15), AKT1 (3), ERBB2 (17-21, 24, 26), NRAS (2-4), PIK3CA (9, 20)

   Colon Cancer
   BRAF (15), KRAS (2- 4), NRAS (2-4)

   GIST
   KIT (8-11, 13, 14, 17, 18) PDGFRA (12, 14, 18), BRAF (15)

   Melanoma
   BRAF (15), GNA11 (5), GNAQ (5), KIT (8-11, 13, 14, 17, 18), NRAS (2-4)

   Thyroid Cancer
   BRAF (15), KRAS (2-4), NRAS (2-4), RET (16)

2. Genes sequenced by the **Illumina TruSight Myeloid panel** (if only part of a gene is sequenced, the exons are indicated in brackets):

   ABL1 (4-6), ASXL1 (12), ATRX (8-10, 17-31) BCOR, BCORL1, BRAF (15), CALR (9), CBL (8+9), CBLB (9, 10), CBLC (9, 10), CDKN2A, CEBPA, CSF3R (14-17), CUX1, DNMT3A, ETV6/TEL, EZH2, FBXW7 (9-11), FLT3 (14+15+20), GATA1 (2), GATA2 (2-6), GNAS (8+9), HRAS (2+3), IDH1 (4), IDH2 (4), IKZF1, JAK2 (12+14), JAK3 (13), KDM6A, KIT (2, 8-11, 13+17), KRAS (2+3), MLL (5-8), MPL (10), MYD88 (3-5), NOTCH1 (26-28,+34), NPM1 (12), NRAS (2+3), PDGFRA (12+14+18), PHF6, PTEN (5+7), PTPN11 (3+13), PAR21, RUNX1, SETBP1 (4), SF3B1 (13-16), SMC1A (2, 11, 16+17), SMC3 (10, 13, 19, 23, 25,+28), SRSF2 (1), STAG2, TET2 (3-11), TP53 (2-11), U2AF1 (2+6), WT1 (7+9), ZRSR2

   Genes analyzed for:

   Myeloproliferative neoplasms

   CALR (9, 10), JAK2 (12+14) and MPL (10)

   Lymphoma

   MYD88 (3-5)

   Myelodysplastic syndrome with ringed sideroblasts

   SF3B1 (13-16)

   Acute Myeloid Leukemia

   DNMT3A,*FLT3 (14+15+20), IDH2 (4), KIT (2, 8-11, 13, 17), *NPM1 (12), CEBPA, TP53 (2-11), SRSF2 (1), ASXL1 (12)

   *FLT3 and NPM1 will continue to be done in parallel, with fragment analysis technology

<u>Specimen/information required:</u>

Hematologic specimens:

- A bone marrow sample collected in EDTA
- Lymph node (5mm^3 fresh tissue in sterile medium or frozen preferred)

Solid Tumor Specimens:

- Paraffin embedded tissue
  - block with tumor
- Fresh tissue
  - 5mm^3 in sterile medium or frozen

Analysis for fusion genes such as ALK, PML-RARa and BCR-ABL, will continue to be done using current methods until these assays are validated by NGS.

If you have any questions please contact the Molecular Diagnostic Lab at 902-473-6665, Dr Wenda Greer at 902-473-6691 (wenda.greer@nshealth.ca), Dr. Weei Huang for solid tumor questions at 902-473-7665 (weei-yuan.huang@nshealth.ca) or Faye Lively at 902-473-6387 (faye.lively@nshealth.ca)