INSELSPITAL
Universitätsklinik für Hämatologie und
Hämatologisches Zentrallabor

NEWSLETTER MARCH 2017

HEMATOLOGIC MOLECULAR DIAGNOSTICS

Order form

Kindly use the new order form in German or French on our Website

www.hzl.insel.ch/de/labor/haematologische-molekulare-diagnostik

The form can be filled online, printed and shipped with the sample. Shipping details are listed on the order form. Paper forms can be ordered at the laboratory (Tel +41 31 632 03 09).

Preserving of samples

In case of unclear diagnosis, samples can be sent for preservation. Analyses can be ordered anytime.

Further analyses

For further analyses and questions, please contact +41 31 632 03 09 or e-mail to elisabeth.oppliger@insel.ch or naomi.porret@insel.ch

NEW ANALYSES

ANALYSE	GENE
AML Diagnostic Panel	AML1-ETO (RUNX1-RUNX1T1), BCR-ABL1, CBFB-MYH11, PML-RARA, EVI1, ASXL1, CEBPA, EZH2, FLT3 ITD, IDH1/2, NPM1, RUNX1, TP53
AML Diagnostic Panel small	AML1-ETO (RUNX1-RUNX1T1), CBFB-MYH11, FLT3 ITD, NPM1
AML Mutation Panel (19 genes)	ASXL1, BRAF, CBL, CEBPA, DNMT3A, FLT3, GATA2, IDH1/2, JAK2, KIT, KRAS, NPM1, NRAS, PTPN11, RUNX1, TET2, TP53, WT1
AML Relapse	AML1-ETO (RUNX1-RUNX1T1), CBFB-MYH11, PML-RARA, EVI1, ASXL1, CEBPA, EZH2, FLT3 ITD, IDH1/2, NPM1, RUNX1, TP53
MDS Prognostic Panel	ASXL1, EZH2, RUNX1, TP53
MDS Mutation Panel (20 genes)	ASXL1, CBL, CEBPA, DNMT3A, ETV6, EZH2, GATA2, IDH1/2, JAK2, KRAS, NRAS, RUNX1, SETBP1, SF3B1, SRSF2, TET2, TP53, U2AF1, ZRSR2
MDS RARS, SF3B1	SF3B1
CMML Panel	ASXL1, CBL, EZH2, JAK2, KRAS, NRAS, RUNX1, SETBP1, SRSF2, TET2
MPN Mutation Panel	ASXL1, CBL, DNMT3A, EZH2, IDH1/2, JAK2, MPL, SF3B1, SRSF2, SOCS1, TET2, TP53, U2AF1, ZRSR2
Mastocytosis Panel	ASXL1, RUNX1, SRSF2
Monitoring	Individual therapy monitoring by mutation analysis

AML

The **AML Diagnostic Panel** includes the 4 translocations AML1-ETO (RUNX1-RUNX1T1), BCR-ABL1, CBFB-MYH11 und PML-RARA, EVI1 expression and screening for mutations in 9 relevant genes. NPM1 mutations and biallelic CEBPA mutations are associated with a favorable prognosis while mutations in ASXL1, EZH2, FLT3, RUNX1, TP53 and overexpression of EVI1 have a poor prognosis. Mutations in IDH1/2 und FLT3 are targets for targeted therapy with IDH- and FLT3-inhibitors.

In addition, the comprehensive **AML Mutation Panel** including 19 genes and an **AML Relapse Panel** are available. Single mutations detected at diagnosis can be ordered for follow-up.

Shipping of samples with Mondexpress

For shipping of samples for RNA extraction, use Mondexpress of the Swiss post. The material should be shipped in the afternoon. Delivery the next morning until 9 h is guaranteed. Costs CHF 16–20.

In case of sampling for RNA extraction (marked with Express1) on the order form) on Friday and weekends, please use 2 PAXgene tubes for blood or bone marrow to preserve the RNA. Tubes can be ordered in the lab. For combined orders an additional blood or BM tube is required.

Impressum

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MDS

Somatic mutations in ASXL1, DNMT3A, EZH2, RUNX1, SF3B1, SRSF2, TET2, TP53 und U2AF1 confirm a clonal hematopoiesis. The **MDS Prognostic Panel** includes the 4 genes ASXL1, EZH2, RUNX1 und TP53 associated with a particularly poor prognosis. The **MDS Mutation Panel** comprises 20 genes including the spliceosome genes. In case of low risk MDS and CHIP a reimbursement confirmation should be requested since this analysis is not a standard benefit at the moment.

The SF3B1 mutation can be ordered separately. According to the WHO classification 2016, the mutation confirms a MDS RARS in the presence of only 5% ringsideroblasts.

Bone marrow samples can be sent for preservation. Subsequent analyses can be ordered anytime.

CMML

About 90% of CMML patients carry somatic mutations. The most frequently mutated genes are TET2 (~60%), SRSF2 (~50%), ASXL1 (~40%), and NRAS (~30%). Mutations in ASXL1, CBL, EZH2, NRAS, RUNX1, SETBP1 und SRSF2 are associated with shorter survival.

MYELOPROLIFERATIVE NEOPLASIA

JAK2, CALR und MPL mutations are part of the diagnostic criteria for MPN. According to the WHO 2016, triple negative patients with PMF or pre-PMF should be screened for mutations in ASXL1, EZH2, IDH1/2, SF3B1, SRSF2 and TET2 to determine the clonal nature of the disease. ASXL1 mutations are risk factors for transformation into MF and AML.

The **MPN Mutation Panel** comprises 15 additional genes including the whole coding region of JAK2 (incl. exon 12-mutations) und MPL. CALR has to be ordered separately.

MASTOCYTOSIS

In c-kit positive systemic mastocytosis, mutations in ASXL1, RUNX1 und SRSF2 are associated with poor prognosis.

MYELOID NEOPLASIA WITH GERMLINE PREDISPOSITION (WHO 2016)

Myeloid neoplasia with germline predisposition is a new entity in the WHO 2016. A gene panel is in preparation.

TP53

In CLL, TP53 mutations and/or deletions of 17p are associated with poor prognosis. These patients might respond favorably to the new B-cell receptor- and BCL2-inhibitors. Accordingly, TP53 analysis is recommended for all patients before start of therapy. The analysis by NGS allows detection of small clones at a sensitivity of >5%.

In myeloid neoplasia (AML, MDS, MDS with del5q and MPN), TP53 mutations are associated poor prognosis, progression and transformation.

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