GENOMED
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REQUEST FORM
- HEMATOLOGY AND HEMATO-ONCOLOGY

Please Post label(s) and/or FILL:

PATIENT IDENTIFICATION:

*Name: *Gender: F ☐ M ☐

*D.B.: / / Identification Number:

Adress:

E-mail: Telephone:

*Mandatory

REFFERNG DOCTOR

***Name:** ***ID number.:**

***Hospital / Service:** **Telephone:**

Do you authorize the report to sent by e-mail? Yes ☐ No ☐
If yes, please write your **institutional email** address :

***Informed Consent** (to be filled by the referring doctor):

I hereby declare that the patient informed consent for diagnosis was obtained. Yes ☐ No ☐

I hereby declare that the patient informed consent for investigation was obtained. Yes ☐ No ☐

SPECIMEN DETAILS

Peripheral blood ☐ Bone Marrow ☐

Lymph Node ☐ Other ☐_____

Collection date and time:
____/____/____ _____:_____h

*CLINICAL DATA

Diagnosis:

Clinical Situation: Initial diagnosis ☐ Relapse ☐ Monitoring ☐

Therapy: No ☐ Yes ☐ Which?_____

CYTOGENETICS STUDIES

Conventional cytogenetics (Peripheral blood / Bone Marrow in heparin) – SNS codes in blue

☐ 34100 Karyotype (cell culture and analysis)

☐ 34025 Cell culture and analysis

Molecular cytogenetics - FISH (Peripheral blood / Bone Marrow in heparin or EDTA)

☐ 34900 Processing for FISH studies

*Paraffin embedded tissue sections (3x50 µm): code 31710

PANELS:

Chronic Lymphocytic Leukemia
☐ 5x34156* Panel 1 - 11q-, 17p-, +12, IgH, 13q-
☐ 7x34156* Panel 2 - 11q-, 17p-, +12, IgH, 13q- (D13S319 e D13S25), 6q-

Mielodysplastic Syndromes
☐ 3x34156 Panel 1 - 5q-, 7q-, 20q-
☐ 5x34156 Panel 2 - 5q-, 7q-, 20q-, +8, 17p-
☐ 34301 Panel 3 - 5q- (5q31 e 5q33-34), 7q-, 20q

Acute Myeloid Leukemia
☐ 4x34156 t(15;17), t(8;21), *KMT2A*, inv(16)

Chronic Myeloid Leukemia
☐ 34156 t(9;22)

Multiple Myeloma
☐ 36229 Panel 1 - clg-FISH: 13q-, 17p-, t(4;14), t(11;14), t(14;16)
☐ 7x34156 Panel 2 - clg-FISH: 13q-, 17p-, t(4;14), t(11;14), t(14;16), 1q+, aneuploidies 5, 9 e 15
☐ 4x34156 Panel 3 - clg-FISH: 17p-, t(4;14), t(14;16), 1q+

Diffuse Large Cells Lymphoma
☐ 3x34156* t(14;18), *MYC* e *BCL6*

Follicular Lymphoma
☐ 34156* t(14;18)

Burkitt Lymphoma
☐ 34156* t(8;14)

Malt Lymphoma
☐ 2x34156* t(11;18) e t(14;18) *IgH::Malt1*

Mantle Cell Lymphoma
☐ 34156* t(11;14)

Neoplasms with Eosinophilia
☐ 34156 *PDGFR β*
☐ 34156 *PDGFR α*
☐ 34156 *FGFR1*

Signature of the referring doctor: _____ **Date** ___/___/___

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SPECIFIC STUDIES:

- ☐ 34156* t(14q32) *IgH*
- ☐ 34156* t(2p23) *ALK*
- ☐ 34156* t(8q24) *MYC*
- ☐ 34156* t(3q27) *BCL6*
- ☐ 34156* t(8;14)(q24;q32) *IgH::MYC*
- ☐ 34156* t (11;14)(q13;q32) *IgH::CCND1*
- ☐ 34156* t(14;18)(q32;q21) *IgH::BCL2*
- ☐ 34156* t(14;18)(q32;q21) *IgH::Malt1*
- ☐ 34156* t(11;18)(q21;q21) *BIRC3::Malt1*
- ☐ 34156 t(14;16)(q32;q23) *IgH::MAF*
- ☐ 34156 t(4;14)(p16;q32) *IgH::FGFR3*
- ☐ 34156 Aneuploidy of 5, 9 and 15
- ☐ 34156 1q+
- ☐ 34156* 17p- [del(17p13) TP53]
- ☐ 34156* 6q- [del(6q21)]
- ☐ 34156* cenX/cenY
- ☐ 34156* t(17q12-q21) *RARA*
- ☐ 34156 t(15;17)(q22;q21) *PML::RARA*
- ☐ 34156 inv(16)/t(16;16))(p13;q22)
- ☐ 34156 t(9;22)(q34;q11.2) *BCR::ABL1*
- ☐ 34156 t(8;21)(q22;q22) *RUNX1::RUNX1T1*
- ☐ 34156 t(11q23) KMT2A
- ☐ 34156 *PDGFR α*
- ☐ 34156 *PDGFR β*
- ☐ 34156 *FGFR1*
- ☐ 34156* 11q- [del(11q22.3) *ATM*]
- ☐ 34156* 13q- [del(13q14.3) D13S319]
- ☐ 34156* 13q- [del(13q14.3) D13S25]
- ☐ 34156 -5 ou 5q- [del(5q31)]
- ☐ 34156 -5 ou 5q- [del(5q33-34)]
- ☐ 34156 -7 ou 7q- [del(7q31)]
- ☐ 34156 20q- [del(20q12)
- ☐ 34156 cen 8
- ☐ 34156 +12 [cen12]
- ☐ 34156 .....

MOLECULAR BIOLOGY STUDIES (Peripheral blood / Bone Marrow in EDTA) – SNS codes in blue

- ☐ 34201 DNA Extraction
- ☐ 34205 RNA Extraction

Fusion gene transcripts:

- ☐ 34412 t(8;21) *RUNX1::RUNX1T1*
- ☐ 34409 t(15;17) *PML::RARA*
- ☐ 34584 inv(16) *CBFB::MYH11*
- ☐ 34209 t(4;11) *KMT2A::AFF1*
- ☐ 34403 t(9;22) *BCR::ABL1*
- ☐ 34210 del(1) *SIL::TAL1*
- ☐ 34622 t(12;21) *ETV6::RUNX1*

Fusion transcripts Quantification:

- ☐ 36219 t(9;22) *BCR::ABL1 (p190)*
- ☐ 36219 t(9;22) *BCR::ABL1 (p210)*
- ☐ 36219 t(15;17) *PML::RARA (bcr1)*
- ☐ 36219 t(15;17) *PML::RARA (bcr3)*

Mutation assays:

- ☐ 36220 *BCR::ABL1* resistance mutations
- ☐ 36214 *FLT3* (ITD e TDK)
- ☐ 36215 *NPM1* (exão 12)
- ☐ 34900 *CEBPA*
- ☐ 34900 *RUNX1*
- ☐ 34900 *IDH1* (exon 4)
- ☐ 34900 *IDH2* (exon 4)
- ☐ 34900 *ASXL1* (exons 12 and 13)
- ☐ 34847 *KIT* (D816V)
- ☐ 36250 *JAK2* (V617F)
- ☐ 34900 *CALR* (exon 9)
- ☐ 36245 *MPL* (W515L/K/S/A)
- ☐ 36251 *JAK2* (exon 12)
- ☐ 34900 *MYD88* (L265P)
- ☐ 34900 *CXCR4* (c-terminal)
- ☐ 36314 *BRAF* (V600E)
- ☐ 34900 *TP53*
- ☐ 36168 mutational status of *IGHV* genes

Other Studies

- ☐ 34900 .....

NGS Panels

- ☐ 34900 **Mieloyd Panel** (30 genes: *ABL1, ASXL1, BRAF, CALR, CBL, CEBPA, CSF3R, DNMT3A, ETV6, EZH2, FLT3, HRAS, IDH1, IDH2, JAK2, KIT, KRAS, MPL, NPM1, NRAS, PTPN11, RUNX1, SETBP1, SF3B1, SRSF2, TET2, TP53, U2AF1, WT1, ZRSR2)*
- ☐ 34900 **Acute Myeloid Leukemia** (20 genes: *ASXL1, BRAF, CEBPA, DNMT3A, EZH2, FLT3, IDH1, IDH2, KIT, KRAS, NPM1, NRAS, RUNX1, SF3B1, SRSF2, TET2, TP53, U2AF1, WT1, ZRSR2)*
- ☐ 34900 **Mielodysplastic Syndromes** (24 genes: *ASXL1, BRAF, CBL, CEBPA, CSF3R, DNMT3A, ETV6, EZH2, FLT3, HRAS, IDH1, IDH2, KRAS, MPL, NPM1, NRAS, RUNX1, SF3B1, SRSF2, TET2, TP53, U2AF1, WT1, ZRSR2)*
- ☐ 34900 **Myeloproliferative Neoplasms Mieloproliferativos** (10 genes: *ASXL1, CALR, EZH2, IDH1, IDH2, JAK2, MPL, SRSF2, TP53, U2AF1)*
- ☐ 34900 **Chronic Myelomonocytic Leukemia** (8 genes: *ASXL1, CBL, KRAS, NRAS, RUNX1, SETBP1, SRSF2 e TET2)*
- ☐ 34900 **Juvenile Myelomonocytic Leukemia** (7 genes: *CBL, KRAS, NRAS, PTPN11, RUNX1, SETBP1, ZRSR2)*

Chimerism

- ☐ 34425 Pre-transplant
- ☐ 34425 Post-transplant

Clonality Assays

- ☐ 36166 B Clonality assay - IGH
- ☐ 36166 B Clonality assay - IGK
- ☐ 36167 T Clonality assay - TCRB
- ☐ 36167 T Clonality assay - TCRG

Hematology (other tests)

- ☐ 34301 Antithrombin III deficiency: *SERPINC1* gene
- ☐ 34497 Trombophilias Panel - Factor II, Factor V, *MTHFR* and *PAI1*
- ☐ 34370 Factor II deficiency (*F2* gene; variant G20210A)
- ☐ 34361 Factor V deficiency (*F5* gene): Leiden mutation
- ☐ 34367 Hyperhomocysteinemia: *MTHFR* gene (C677T e A1298C)
- ☐ 34364 *PAI1* gene (4G mutation)
- ☐ 34310 Factor VII deficiency: *F7* gene - index case
- ☐ 34900 Factor XII deficiency: *F12* gene (C46T mutation)
- ☐ 34305 Protein S deficiency: *PROS1* gene - index case
- ☐ 34900 Osler-Weber-Rendu disease: *ACVRL1* gene
- ☐ 34900 Osler-Weber-Rendu disease: *ENG* gene
- ☐ 34900 Osler-Weber-Rendu disease: *ACVRL1* and *ENG* genes (MLPA)
- ☐ 36168 von Willebrand disease type 1, 2, 3: *VWF* gene
- ☐ 36168 Sickle Cell Anemia: *HBB* gene
- ☐ 36190 Haemocromatosys (H63D, C282Y and S65C mutatioons: *HFE* gene)
- ☐ 34837 Gilbert Syndrome:TA Insertion - *UGT1A1* gene (level I)
- ☐ 34838 Gilbert/ Crigler Najjar Syndrome: *UGT1A1* gene (level II)
- ☐ 34900 Familial Congenital neutropenia: *ELA2 gene*

Signature of the referring doctor: _____ **Date** ___/___/___

SAMPLE COLLECTION, PRESERVATION AND SHIPPING

SAMPLE TYPE / STUDY	Peripheral blood / Bone Marrow in heparin tube	Peripheral blood / Bone Marrow in EDTA tube	Paraffin	Lymph Node
Karyotype	X			X
FISH	X	X	X	X
Molecular Biology		X	X	X
Quantity	2 - 5 mL	5 - 10 mL	**FISH**: Paraffin block+ 1 H&E slide with tumour tissue defined or 3 sections of 50 µm **Molecular**: 6 sections of 10µm; 3 sections for eppendorf	1 (fragment)
Sample stability and shipping conditions	Room temperature (24-48h; preferably 24h)	Room temperature (24-48h; preferably 24h)	Room temperature	Room temperature, in sterile recipient with culture media or saline solution (24-48h; preferably 24h)

Notes: - *Collection material to be provided by the Laboratory, upon request.*

Informed Consent (mandatory - to be filled by the patient)

I hereby declare that I have read the Privacy Policy of GenoMed® - Diagnósticos de Medicina Molecular, S.A., available at https://genomed.pt/politica-de-privacidade-e-de-cookies/ and I give my consent to the processing of the personal data.

☐ Agree ☐ Not agree

Hereby I _____ [name], _____/_____/_____ [date of birth], give my consent that my / my child´s biological sample will be examined for genetic changes (mutations) in the specified gene(s), related to the diseases / clinical features described above. Herewith I declare that I have been informed about the chances and limitations of the requested testing procedure. I was informed in detail about the consequences resulting from the test results. I agree that the sample may be stored in order to allow repetition of the tests or further related tests in the future. All data about me / my child are subject to medical confidentiality. They can be disclosed to family members or their physicians only with my permission, but not to third parties. I am entitled to revoke this consent at any time.

☐ Agree ☐ Not agree

I agree that my / my child´s tests results and/or clinical data may be used in scientific publications in anonymized form in case of approval of the Ethics Committee .

☐ Agree ☐ Not agree

(According to the Direction of General Health updated standard 015/2013)

Patient's Signature: Date: / /

Signature of the referring doctor: _____ Date ___/___/___