

<i>Landscape</i>	<i>Description</i>	<i>Sample type</i>	<i>Amount*</i>	<i>Maximum turnaround time (days)</i>
PCA3/PSA score				
PCA3 (Prostate cancer antigen 3) is a fragment of non-coding RNA that is overexpressed in prostate cancer cells. It is detectable in prostate cells that are shed in the urine after a digital rectal exam (DRE). The assessment of PCA3 levels in urine is a method for early detection and diagnosis of prostate cancer, and has the potential to reduce unnecessary prostate biopsies.	Determination of the PCA3/PSA score by qRT-PCR.	First urine after DRE.	20-30 ml (Stock at 4 ° C and process into 4 h).	7
		RNA	1 µg	
c-MET gene amplification				
C-MET gene is activated in different types of tumours through its amplification at the genomic level. This activation has been associated with the prognosis of these tumours and the prediction of the response to MET inhibitors.	Quantification, by qRT-PCR of the amplification of the intron10-exon11 junction of the c-MET gene.	FFPE tissue.	Block or sections	7
		Frozen tissue.	As much amount as possible	
		DNA	2 µg	
t(9;22) or BCR-ABL fusion gene				
t (9; 22)(q34;q11) translocation is present in practically all cases of chronic myeloid leukaemia (95%) and in some cases of pre-B type acute lymphocytic leukaemia, being associated with aggressive disease and poor prognosis. This assay is useful for the diagnosis and determination of minimal residual disease during the treatment.	Quantification of the levels of p190 or p210 variants of the BCR-ABL fusion gene by qRT-PCR.	Peripheral blood.	5 ml	14
		Bone marrow aspirate.	2 ml	
		RNA.	2 µg	
t(8;21) or AML1-ETO fusion gene				
t(8;21)(q22;q22) translocation is one of the most frequent chromosomal alterations in acute leukaemias (especially in the M2 subtype) and a good prognostic marker. The test is used for the diagnosis and determination of minimal residual disease after treatment.	Detection of the AML1-ETO fusion gene by qRT-PCR.	Peripheral blood.	5 ml	14
		Bone marrow aspirate.	2 ml	
		RNA	2µg	
t(15;17) or PML-RARα fusion gene				
t(15;17)(q24;q21) translocation is associated with acute promyelocytic leukaemia (M3 type) as a biomarker of good prognosis. This assay is used for diagnosis and determination of minimal residual disease after treatment.	Detection of the bcr1, bcr2 y bcr3 variants of the PML-RARα fusion gene by qRT-PCR.	Peripheral blood.	5 ml	14
		Bone marrow aspirate.	2 ml	
		RNA.	2 µg	

inv(16) or MYH11- CBFβ fusion gene				
inv(16)(p13q22) is associated with acute myeloid leukemia with abnormal bone marrow eosinophilia (AML-M4Eo subtype) as a biomarker of good prognosis. This assay is used for diagnosis and determination of minimal residual disease after treatment.	Detection and quantification of the levels of A, D and E variants of the chromosome 16 inversion using qRT-PCR.	Peripheral blood.	5 ml	14
		Bone marrow aspirate.	2 ml	
		RNA.	2 µg	
t(14;18) or BCL2-IgH fusion gene				
t(14;18)(q32;q21) translocation is associated with the diagnosis of follicular lymphoma and some cases of large B cell lymphomas. This test is used for the detection of minimal residual disease after treatment. The size of the amplified sequence is normally used to demonstrate the clonal identity between pre and post-treatment samples from the same patient. 60% of the cases present the MBR (major breakpoint region) variant and 5-10% the mcr (minor cluster region) variant.	Quantification of the levels of MBR or mcr variants of the BCL2-IGH fusion gene by qRT-PCR.	Peripheral blood.	5 ml	14
		Bone marrow aspirate.	2 ml	
		FFPE tissue.	Block or sections	
		Frozen tissue.	As much amount as possible	
		DNA	2 µg	
t(11;14) or BCL1-IgH fusion gene				
Although the t(11;14)(q13;q32) translocation is present in other lymphoproliferative diseases, it occurs mainly in mantle cell lymphomas (50-70%), which are more aggressive and have, in general, a worse prognosis than other low-grade B-cell lymphomas. This test is used not only to diagnose patients with a suspected mantle cell lymphoma, but also to monitor and detect relapse of the disease.	Determination of the presence of the BCL1-IgH fusion gene by end-point PCR.	Peripheral blood.	5 ml	7
		Bone marrow aspirate.	2 ml	
		FFPE tissue.	Block or sections	
		Frozen tissue.	As much amount as possible	
		DNA	2 µg	
Ig H and K genes rearrangements				
The presence of clonal rearrangements of the IgH and IgK genes is suggestive of the presence of a leukemia or a lymphoma of B-cell. This test is therefore useful to support its diagnosis by histopathological and/or flow cytometry techniques.	Determination of clonality in IgH (FR1, FR2, FR3, DH-JH and DH7-JH regions) and IgK (Vκ-Jκ and Vκ-Kde + intron-Kde regions) genes by fluorescence-labelled PCR and capillary electrophoresis.	Peripheral blood.	5 ml	14
		Bone marrow aspirate.	2 ml	
		FFPE tissue.	Block or sections	
		Frozen tissue.	As much amount as possible	
		DNA	5 µg	
TCR β and γ genes rearrangements				
The presence of clonal rearrangements of TCRβ and TCRγ genes is suggestive of the presence of a leukemia or a lymphoma of T-cell. This test is therefore useful to support its diagnosis by histopathological and/or flow cytometry techniques.	Determination of clonality in TCRβ (Vβ + Jβ1/2, Vβ + Jβ2 and Dβ + Jβ1/2 regions) and TCRγ (All V and J genes, Vγ9, Vγ10 and Jγ1/Jγ2 regions) by fluorescence-labelled PCR and capillary electrophoresis.	Peripheral blood.	5 ml	14
		Bone marrow aspirate.	2 ml	
		FFPE tissue.	Block or sections	
		Frozen tissue.	As much amount as possible	
		DNA	5 µg	
Microsatellite instability				
The existence of genome instability in gastric, colon or endometrial	Determination by multiplex	Peripheral blood.	5 ml	14

cancer represents a predictor of the therapeutic efficacy of adjuvant chemotherapy with fluorouracil.	fluorescent PCR and capillary electrophoresis of BAT25 and BAT26 markers (recommended by NCI) and NR21, NR22 and NR24 monomorphic markers.	Bone marrow aspirate.	2 ml	
		FFPE tissue.	Block or sections	
		Frozen tissue.	As much amount as possible	
		DNA	2 µg	
Mutations in BCR-ABL				
The presence of mutations in the kinase domain of the BCR-ABL fusion gene is associated with the resistance of patients by not responding to treatment and relapsing.	Detection of mutations in the full tyrosine kinase domain of the BCR-ABL gene by direct sequencing.	Peripheral blood.	5 ml	14
		Bone marrow aspirate.	2 ml	
		RNA total	2 µg	
Mutations in JAK2				
Detection of mutations in the JAK2 kinase, especially V617F mutation in exon 14, is indicated in patients with suspected chronic Philadelphia negative myeloproliferative neoplasms (polycythaemia vera, essential thrombocythemia and primary myelofibrosis) since they have been shown to have a higher frequency of complications (secondary fibrosis, haemorrhages and thrombocytosis) and that they need cytoreductive treatment more frequently than patients with the native JAK2 gene.	Detection of mutations in exons 12 and 14 of the JAK2 gene by Sanger sequencing. Determination of the V617F (JAK2) allelic burden by qRT-PCR.	Peripheral blood.	5 ml	14
		Bone marrow aspirate.	2 ml	
		DNA	2 µg	
Mutations in MPL				
Mutations in exon 10 of the MPL gene, exclusive of mutations in the JAK2 gene, are found in approximately 3-4% of patients with essential thrombocythemia (ET) and 4-8% with myelofibrosis. The MPL gene encodes the thrombopoietin receptor, which regulates the differentiation of megakaryocytes and platelets. Patients with mutations in MPL have been shown to be more anaemic than those without the mutation. Patients with ET and mutations in MPL have a higher platelet count and isolated megakaryocytic proliferation than those with the V617F mutation in JAK2.	Detection of mutations in exon 10 of MPL gene by direct sequencing.	Peripheral blood.	5ml	14
		Bone marrow aspirate.	2ml	
		DNA	2 µg	
Mutations in CALR				
Mutations in the CALR (calreticulin) gene allows the diagnosis of patients with essential thrombocythemia or primary myelofibrosis who have proved to be negative in the detection of mutations in the JAK2 gene.	Detection of mutations in exon 9 of CALR gene by direct sequencing.	Peripheral blood.	5ml	14
		Bone marrow aspirate.	2ml	
		DNA	2 µg	
Mutations in TET2.				
Mutations in the tumour suppressor gene TET2 allows to complement the diagnosis of patients with myeloproliferative neoplasms (MPNs). Mutations in this gene (13% of MPNs) cause genetic instability through epigenetic modifications and promote cancer progression. The sequence in which these mutations are acquired is critical. Therefore, patients with early mutations in TET2 were more likely to have a better prognosis compared to patients who had previous mutations in other genes related to MPNs.	Detection of mutations in exons 3, 6, 7, 9, 10 and 11 of TET2 gene by direct sequencing.	Peripheral blood.	5ml	21
		Bone marrow aspirate.	2ml	
		DNA.	2 µg	

Mutations in NPM1				
Mutations in nucleophosmin (NPM1) gene have been detected in about 50% of Acute Myeloid Leukemia (AML) adults with a normal karyotype. Their presence associated with the absence of mutations (tandem internal duplications) in the FLT3 gene predicts a favorable outcome in these patients.	Detection of mutations in exon 12 of NPM1 gene by direct sequencing.	Peripheral blood.	5ml	14
		Bone marrow aspirate.	2ml	
		DNA		
Mutations in FLT3				
Internal tandem duplications (ITD) in the FLT3 gene in acute myeloid leukaemias are indicative of poor prognosis. In addition, patients with a high FLT3-ITD allele burden (> 0.5) and linked with a mutated MPN1 are considered of high risk and thus indicated for HSCT (Hemopoietic Stem Cell Transplant).	Detection of ITDs and D835 mutation in the FLT3 gene by fluorescence-labelled PCR, restriction analysis and capillary electrophoresis.	Peripheral blood.	5 ml	14
		Bone marrow aspirate.	2 ml	
		DNA	2 µg	
Mutations in WT1				
Patients with acute myeloid leukemia (AML) have been shown to form a heterogeneous group of neoplasms. Their appropriate care depends on their cytogenetic characterization, which stratifies them into three large groups correlated with a good, intermediate or bad prognosis. In the intermediate risk group (40-50% of patients with AML), who lack an altered karyotype, the detection of mutations in exons 7 or 9 of the WT1 (Wilms tumour 1) gene plus an internal repetition in tandem in the FLT3 gene have been linked to a worse clinical course of younger adult patients.	Detection of mutations in exons 7 and 9 of WT1 gene by direct sequencing.	Peripheral blood.	5ml	14
		Bone marrow aspirate.	2ml	
		DNA	2 µg	
Mutations in IDH1 & IDH2				
Patients with acute myeloid leukemia (AML) have been shown to form a heterogeneous group of neoplasms. Their appropriate care depends on their cytogenetic characterization, which stratifies them into three large groups correlated with a good, intermediate or bad prognosis. In the intermediate risk group (40-50% of patients with AML), without an altered karyotype, detection of mutations in the gene gene in exon 4 of the IDH1 and/or IDH2 genes (isocitrate dehydrogenases 1 and 2) are good prognostic markers in patients with a mutated NPM1 gene and native FLT3. Mutations in the IDH1 and/or 2 gene are also useful to support the diagnosis of low-grade gliomas and secondary glioblastoma.	Detection of mutations in exon 4 of IDH1 e IDH2 genes by direct sequencing.	Peripheral blood.	5ml	14
		Bone marrow aspirate.	2ml	
		DNA	2 µg	
Mutations in CEBPA				
Patients with acute myeloid leukemia (AML) have been shown to form a heterogeneous group of neoplasms. Their appropriate care depends on their cytogenetic characterization, which stratifies them into three large groups correlated with a good, intermediate or bad prognosis. In the intermediate risk group (40-50% of patients with AML), who lack an altered karyotype, the detection of biallelic mutations in the CEBPA (CCAAT / enhancer binding protein alpha) gene are indicative of a	Detection of mutations in whole CEBPA gene by direct sequencing.	Peripheral blood.	5ml	21
		Bone marrow aspirate.	2ml	
		DNA	2 µg	

good prognosis in patients with normal cytogenetics and native NPM1 and FLT3 genes.				
Mutations in GATA1				
Somatic mutations in GATA1 gene allow to detect an increased risk of developing acute megakaryoblastic leukemia (DE-AML M7) in newborns with Down syndrome.	Detection of mutations in the exons 1 and 2 of the GATA1 gene by direct sequencing.	Peripheral blood.	5 ml	14
		Bone marrow aspirate.	2 ml	
		DNA	2 µg	
Mutations in MYD88				
It is believed that Waldenström's macroglobulinemia (MW) derives from a multiphase transformation process that accumulates sequential oncogenic phenomena in which the most striking is the L265P mutation of the MYD88 gene (~ 90% of the MW) and whose presence would distinguish the MW from other indolent and chronic B-cell lymphoproliferative diseases. In addition, the mutation may be predictive of patients' sensitivity / resistance to ibrutinib in the activated B cell subtype of DLBCL (diffuse large B-cell lymphoma). Other mutations (V217F, S219C, M232T, S243N and T294P) have also been identified in primary DLBCL tissues.	Detection of mutations in exons 4 and 5 of MYD88 gene by direct sequencing.	Peripheral blood.	5ml	14
		Bone marrow aspirate.	2ml	
		DNA	2 µg	
Mutations in CXCR4				
Mutations in the CXCR4 gene (C-X-C motif chemokine receptor 4) are commonly found in association with the L265P mutation in the MYD88 gene. They are associated with primary resistance and the initial absence of response to inhibitors of BTK, PI3K and mTOR. Therefore, this test is useful to aid in the prognosis and therapeutic management of patients with lymphoplasmacytic lymphoma/Waldenström's macroglobulinemia.	Detection of mutations in whole CXCR4 gene by direct sequencing.	Peripheral blood.	5ml	21
		Bone marrow aspirate.	2ml	
		DNA	2 µg	
Mutations in BRAF				
Mutations in the BRAF gene are found in a wide range of cancers (malignant melanoma, hairy cell leukemia, colorectal, ovarian or lung cancers, etc.). Depending on the neoplasm considered, the BRAF mutational status can serve as a diagnostic, prognostic or predictive marker.	Detection of mutations in exons 11 and 15 of the BRAF gene by direct sequencing.	Peripheral blood.	5 ml	14
		Bone marrow aspirate.	2 ml	
		FFPE tissue.	Block or sections	
		Frozen tissue.	As much amount as possible	
		DNA	2 µg	
Mutations in KIT				
Mutations in the KIT oncogene are associated with gastrointestinal tumors (GISTs, 90% of the patients), acute myeloid leukemia (CBF-AMLs) and mastocytosis. Mutations in the extracellular and self-inhibitory domains are associated with a good response to treatment with imatinib. In contrast, mutations in the kinase domains are associated with resistance to treatment with imatinib. In patients with AML or systemic mastocytosis, mutations in exon 17 usually confer a poor prognosis with increased relapse rate and predict poor survival.	Detection of mutations in exons 9, 11, 13 and 17 of KIT gene by direct sequencing.	Peripheral blood.	5 ml	21
		Bone marrow aspirate.	2 ml	
		FFPE tissue.	Block or sections	
		Frozen tissue.	As much amount as possible	
		DNA	2 µg	
Mutations in PDGFRA				

Mutations in the PDGFRA oncogene are associated with pathomorphologically malignant or high-risk (5-8%) gastrointestinal tumors (GISTs), especially in the 40-50% of KIT wild type GISTs. GISTs with PDGFRA mutations (except D842V in exon 18) are good candidates for tyrosine kinase inhibitor treatment and are likely to respond to imatinib therapy. Mutations in exons 12 and 18 are associated with GISTs of gastric origin and epithelioid morphology, whereas mutations in exon 14 are associated with GISTs in children and young adults. PDGFRA mutations also have been described in synovial sarcomas (SSs), malignant peripheral nerve sheath tumors (MPNST) and myeloid and lymphoid neoplasms with eosinophilia.	Detection of mutations in exons 12, 14 and 18 of PDGFRA gene by direct sequencing.	Peripheral blood.	5 ml	14
		Bone marrow aspirate.	2 ml	
		FFPE tissue.	Block or sections	
		Frozen tissue.	As much amount as possible	
		DNA	2 µg	
Mutations in PTEN				
PTEN is a tumor suppressor gene that mediates cell cycle arrest and apoptosis and is one of the most commonly mutated tumor suppressor genes in cancer. Mutations are found in tumors of the central nervous system, endometrium, prostate, and skin. PTEN loss in breast cancer has been associated with poor response to trastuzumab. These and other PTEN-deficient tumors may respond to PI3 kinase inhibitors. Mutations in the PTEN gene are classified under the multiple hamartoma tumor syndrome PTEN (PHTS). Due to the risk of increased malignancies associated with PHTS, genetic testing is recommended for high-risk patients. The increase in the follow-up of the disease and the evaluation of a prophylactic treatment are available for those who carry PTEN mutations.	Detection of mutations in exons 1 to 9 of PTEN gene by direct sequencing.	Peripheral blood.	5ml	21
		Bone marrow aspirate.	2ml	
		FFPE tissue.	Block or sections	
		Frozen tissue.	As much amount as possible	
		DNA	2 µg	
Mutations in RET				
Mutations in exon 16 of the RET proto-oncogene are responsible for multiple endocrine neoplasia type 2 (MEN 2), a rare inherited disease that is associated with the development of medullary thyroid carcinoma and pheochromocytoma. Therefore, this test allows the prognosis of patients at risk before the clinical symptoms of developing this pathology become evident.	Detection of mutations in exon 16 of RET gene by direct sequencing.	Peripheral blood.	5ml	14
		Bone marrow aspirate.	2ml	
		FFPE tissue.	Block or sections	
		Frozen tissue.	As much amount as possible.	
		DNA	2 µg	
Mutations in TP53				
TP53 is the most frequently mutated gene in human cancers. Especially pancreas, skin, esophagus, head/neck and colorectal cancers, where more than a third of patients have mutations in TP53. The detection of mutations in TP53 is a useful prognostic marker and a potential predictor of response to therapy.	Detection of mutations in exons 5 to 8 of TP53 gene by direct sequencing.	Peripheral blood.	5ml	21
		Bone marrow aspirate.	2ml	
		FFPE tissue.	Block or sections	
		Frozen tissue.	As much amount as possible.	
		DNA	2 µg	
Mutations in KRAS				
Mutations in the KRAS gene, mutually exclusive of mutations in the EGFR gene, can predict resistance to anti-EGFR treatments (Cetuximab and panitumumab) in patients with metastatic colorectal cancer refractory to chemotherapy.	Detection of mutations in the exons 2 and 3 of the KRAS gene by direct sequencing.	Peripheral blood.	5ml	14
		Bone marrow aspirate.	2ml	
		FFPE tissue.	Block or sections	
		Frozen tissue.	As much amount as	

			possible	
		DNA	2 µg	
Mutations in NRAS				
Mutations in the NRAS gene are found in melanoma, and colorectal and thyroid cancers. Approximately 80% of the reported mutations are found in codon 61. They help to classify the subtypes of the disease and can guide targeted therapies. For example, a mutation in NRAS can be predictive of the response of the BRAF inhibitor in patients with metastatic melanoma or predict the response of anti-EGFR therapy in metastatic CRC.	Detection of mutations in exons 2 y 3 of NRAS gene by direct sequencing.	Peripheral blood.	5ml	7
		Bone marrow aspirate.	2ml	
		FFPE tissue.	Block or sections	
		Frozen tissue.	As much amount as possible	
		DNA	2 µg	
Mutations in RUNX1				
RUNX1 gene is the most frequent target of chromosomal translocation associated with human leukaemias. Nevertheless, loss-of-function RUNX1 mutations have also been identified in patients with de novo AML. These alterations could coexist, or be mutually exclusive, with mutations in other genes included in assays already available in our portfolio (i.e. DH1, IDH2, FLT3, TET2, NPM1 and CEBPA genes). Consequently, even if patients with RUNX1 mutations were classified in intermediate risk group due to adverse prognostic outcomes (i.e. shorter relapse-free survival), depending on coexisting detected mutations, they could benefit from specific targeted therapies.	Detection of mutations in exons 4, 5, 6 and 8 of RUNX1 gene by direct sequencing.	Peripheral blood.	5ml	14
		Bone marrow aspirate.	2ml	
		FFPE tissue.	Block or sections	
		Frozen tissue.	As much amount as possible	
		DNA	2 µg	
Mutations in SETBP1				
SETBP1 overexpression associates with worse overall survival of elderly AML patients, Besides somatic gain of function mutations of SETBP1 are associated with myeloid leukemic transformation and convey poor prognosis of MDS and CMML patients.	Detection of mutations in exon 12 of SETBP1 gene by direct sequencing.	Peripheral blood.	5ml	7
		Bone marrow aspirate.	2ml	
		FFPE tissue.	Block or sections	
		Frozen tissue.	As much amount as possible	
		DNA	2 µg	
Mutations in ASXL1				
Somatic mutations in ASXL1 gene has been observed, among many other cancers, mostly in MDS, CMML and AML. ASXL1 mutations prognoses high-risk MDSs, acute transformation of CMML and shorter overall survival of patients with AML	Detection of mutations in exons 13 and 14 of ASXL1 gene by direct sequencing.	Peripheral blood.	5ml	7
		Bone marrow aspirate.	2ml	
		FFPE tissue.	Block or sections	
		Frozen tissue.	As much amount as possible	
		DNA	2 µg	
Mutations in SF3B1				
Somatic mutations in SF3B1 gene has been observed, among many other cancers, mostly in MDS, CMML and AML. SF3B1 structural alterations are predictive of a longer overall survival of MDS patients but shorter in de novo AML and chronic lymphocytic leukemia (CLL) patients.	Detection of mutations in exons 14, 15 and 16 of SF3B1 gene by direct sequencing.	Peripheral blood.	5ml	14
		Bone marrow aspirate.	2ml	
		FFPE tissue.	Block or sections	
		Frozen tissue.	As much amount as possible	

		DNA	2 µg	
Mutations in CSF3R				
Activating mutations in CSF3R are common in patients with chronic neutrophilic leukemia (CNL) or atypical (BCR-ABL1–negative) chronic myeloid leukemia (CML). They represent a useful diagnostic marker and can be also used as predictive marker since patients had showed marked clinical improvement after the administration of the JAK1/2 inhibitor ruxolitinib.	Detection of mutations in exons 14 and 17 of CSF3R gene by direct sequencing.	Peripheral blood.	5ml	14
		Bone marrow aspirate.	2ml	
		FFPE tissue.	Block or sections	
		Frozen tissue.	As much amount as possible	
		DNA	2 µg	
Mutations in EGFR				
Mutations in EGFR gene is associated with a sensitivity to the treatment with tyrosine kinase inhibitors (TKIs) in quimioresistant's patients with non-small cell lung carcinoma (NSCLC). These mutations are therefore predictive markers of therapeutic efficacy and allow the selection of candidates to TKIs therapy.	Detection of mutations in the exons 18,19, 20 and 21 of the EGFR gene by direct sequencing.	Frozen tissue.	As much amount as possible	21
		DNA	2 µg	
		DNA	2 µg	
Mutations in PI3KCA				
Mutations in the PI3K oncogene have been observed in a significant number of cancers, including gastrointestinal, lung and breast cancers. The detection of mutations in this gene is useful to predict the response to therapy of patients.	Detection of mutations in exons 9 y 20 of PI3K gene by direct sequencing.	Frozen tissue.	As much amount as possible	14
		DNA	3 µg	
		DNA	3 µg	
Mutations in AKT1				
AKT1 kinase, a central member of the proliferation and survival pathways, is frequently activated in cancer. A somatic mutation in codon 17 of AKT1 (E17K) has been detected in colorectal, breast, lung and ovarian cancer. The E17K mutation results in the constitutive activation of AKT1 and decreases sensitivity to an allosteric kinase inhibitor. The study of the mutational status of AKT1 in tumors can be used to predict the patient's response to certain therapeutic regimens.	Detection of mutations in exon 4 of AKT1 gene by direct sequencing.	Frozen tissue.	As much amount as possible	14
		DNA	2 µg	
		DNA	2 µg	
Mutations in ERBB2 (HER2)				
Activating mutations in the ERBB2 (also known as HER2) proto-oncogene in patients with breast, ovarian, colorectal, gastroesophageal and pulmonary cancers may cause resistance to treatments with reversible tyrosine kinase inhibitors and, therefore, promote a more aggressive and metastatic disease. Thus, the detection of these mutations significantly broadens the range of patients who could benefit from useful targeted therapies.	Detection of mutations in exons 13, 14 and 22 to 28 of ERBB2 gene by direct sequencing.	Frozen tissue.	As much amount as possible.	21
		DNA	2 µg	
		DNA	2 µg	
Mutations in POLE				
The POLE (Polymerase Epsilon) gene encodes a DNA polymerase with a replication error correction exonuclease activity and, consequently, allows high fidelity replication. A group of endometrial carcinomas have been identified, which are not sufficiently distinctive to allow an accurate diagnosis based on routine histological staining. These tumors are associated with better progression-free survival that	Detection of mutations in exons 9, 13 and 14 of POLE gene by direct sequencing.	Frozen tissue.	As much amount as possible	14
		DNA	2 µg	

does not result from an increased sensitivity to chemotherapy, but is more likely to be linked to increased immunogenicity. Given that clinical practice is to administer adjuvant chemotherapy to the majority of patients with endometrial carcinoma, this test will allow avoiding unnecessary treatments for patients with mutations in POLE.				
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(*) Las cantidades indicadas son cantidades óptimas.

- En sangre periférica para pacientes en tratamiento cuyo número de células sea bajo y para estudios de enfermedad residual se requieren como cantidad óptima 10 ml.
- Para estudios de inestabilidad de microsatélites (MSI) además de tejido tumoral se requieren 3 ml de sangre periférica o tejido normal del mismo paciente.
- Para estudios con tejido fijado se necesitan bloques completos con la biopsia (o 5 secciones de 5 μ m de espesor) que contenga al menos 60% de células tumorales.
- Si se envía una muestra de ADN debe ser ADN genómico.