

ALTUM SEQUENCING, S.L.

Dirección / Address: Avda. Gregorio Peces Barba, 1, 28919, Leganés (Madrid)

Norma de referencia / Reference Standard: **UNE-EN ISO 15189: 2023**

Actividad / Activity: Laboratorio clínico / Medical laboratory

Acreditación / Accreditation nº: **1507/LE2770**

Fecha de entrada en vigor / Coming into effect: 01/09/2023

ALCANCE DE LA ACREDITACIÓN SCHEDULE OF ACCREDITATION

(Rev./ Ed. 2 fecha / Date 07/06/2024)

BIOLOGÍA MOLECULAR / Molecular Biology

ESPECÍMEN / MUESTRA <i>Specimen/Sample</i>	PRUEBAS/ESTUDIOS <i>EXAMINATIONS</i> Método <i>Method</i>	PROCEDIMIENTO <i>PROCEDURE</i>
Sangre Periférica Médula Ósea (EDTA) <i>Peripheral Blood/Bone Marrow (EDTA)</i>	Estudio de variantes somáticas y germinales (SNVs, indels) en genes asociados a patología mielóide mediante secuenciación masiva (NGS) por detección de protones <i>Study of somatic and germline variants (SNVs, indels) in genes associated with myeloid pathology by massive sequencing (NGS) using proton detection</i> <i>Genes with somatic mutations in hematological tumor, epigenetic modifiers: ASXL1, BCOR, BCORL1, DNMT3A, EZH2, IDH1, IDH2, SETBP1 and TET2; myeloproliferative neoplasia driver: CALR; transcription factors: CEBPA, EPAS1, ETV6, GATA1 AND 2, KMT2A, PHF6, RUNX1 and WT1; tyrosine kinase-related genes: CSF3R, EPOR, FLT3, JAK2, KIT, MPL, PTPN11, RIT1 and TYK2; genes related to cohesins: SMC1A, EGLN1 and RAD21; tumor suppressors: KDM6A, NF1, TP53 and VHL; genes related to the anti-apoptosis mTOR pathway: GNB1; genes involved in the production of free radicals in mitochondria: ETNK1; genes involved in the composition of the spliceosome: CUX1, GNAS, SF3A1, SF3B1, SRSF2, STAG2, U2AF1 and ZRSR2. Finally, other genes related to myeloid neoplasms: CBL, KRAS, NPM1, NRAS, RRAS and SH2B3.</i> <i>Genes with germline inherited predisposition mutations involved in bone marrow aplasia: ANKRD26, DDX41, SAMD9 and SAMD9L; genes involved in germline predisposition myeloid neoplasia: ATG2B and GSKIP; mutations in the chromatin of the spliceosome: PRPF8 and STAG2; transcription factors: GATA2 and NFE2; early debut in AML: MBD4; non-driver genes in myeloproliferative neoplasia: PPM1D; genes involved in Noonan syndrome: PTPN11 and RIT1; genes involved in chromosomal maintenance: SMC3; genes in aplastic anemia: TERC and TERT and the gene encoding for thrombopoietin: THPO.</i>	Procedimiento interno <i>Internal procedure</i> SOP6. Ed.8 SOP8. Ed.4

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