

F/D (1)	DISCIPLINE	SPECIMEN	TYPE OF EXAMINATION	DETAILED LIST (2)	METHOD /TECHNICS	TYPE OF PROCEDURE (recognized or internal) (3)	EXAMINATION PROCEDURE (4)	CATEGORY (5)
F	Medical Genetics	Whole blood	Search for known variants	--	PCR + direct Sanger Sequencing	Internal	--	0
D	Medical Genetics	Whole blood	--	Sanger Sequencing - Search for known variants: APC, ATM, BAP1, BARD1, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDKN2A(α e β), CDK4(esone2), CHEK2, CTNNA1, DICER1, FANCM, FH, MC1R, MITF, MLH1, MSH2, MSH3, MSH6, MUTYH, NBN, NF1, NTHL1, PALB2, PIK3CA, PMS2, POLD1, POLE, PTEN, RAD51C, RAD51D, RBT, SMAD4, STK11 e TP53	extraction + PCR + electrophoresis + sequencing	Internal	PI 023 (ed.04,rev.1;2023)	0
F	Medical Genetics	Whole blood	Search for unknown variants	--	Direct NGS Sequencing	Internal	--	0
D	Medical Genetics	Whole blood	--	OncoPan® -Search for unknown germinal variants: ACD, APC, ATM, AXIN2, BAP1, BARD1, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDKN2A(α e β), CDK4(esone2), CHEK2, CTNNA1, EPCAM, FANCM, GREM1, HOXB13, MC1R, MITF(esone9), MLH1, MSH2, MSH3, MSH6, MUTYH, NBN, NTHL1, PALB2, PMS2, POLD1, POLE, POT1, PTEN, RAD51C, RAD51D, RNF43, SMAD4, STK11, TERF21P, TERT e TP53	extraction + NGS	Internal	PI 023 (ed.04,rev.1;2023)	0
D	Medical Genetics	Whole blood	--	OncoPed - Search for unknown germinal variants: APC, BAP1, BRCA1, BRCA2, CDH1, CDKN2A(α e β), CDK4(esone2), DICER1, EPCAM, FBWX7, FH, FLCN, MET, MITF, MLH1, MSH2, MSH6, MUTYH, PALB2, PMS2, POT1, PRKAR1A, PTCH1, PTEN, RB1, SMARCA4, SMARCB1, SUFU, TP53, VHL e WT1.	extraction + NGS	Internal	PI 023 (ed.04,rev.1;2023)	0
F	Medical Genetics	Whole blood	Search for unknown variants	--	MLPA	Recognized	--	0
D	Medical Genetics	Whole blood	--	MLPA - Search for unknown germinal variants: APC, ATM, BARD1, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDKN2(α e β), CDK4, CHEK2, EPCAM, GREM1, MLH1, MSH2, MSH6, MUTYH, PALB2, PMS2, PTEN, TP53, RAD50C, RAD51C, RAD51D, RB1, SMAD4, SMARCB1 e STK11.	extraction + MLPA	Recognized	PI 023 (ed.04,rev.1;2023)	0
F	Medical Genetics	Whole blood	Search for known variants	--	MLPA	Recognized	--	0
D	Medical Genetics	Whole blood	--	MLPA - Search for known variants: APC, ATM, BARD1, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDKN2(α e β), CDK4, CHEK2, EPCAM, GREM1, MLH1, MSH2, MSH6, MUTYH, PALB2, PMS2, PTEN, TP53, RAD50C, RAD51C, RAD51D, RB1, SMAD4, SMARCB1 e STK11.	extraction + MLPA	Recognized	PI 023 (ed.04,rev.1;2023)	0
D	Medical Genetics	Whole blood	--	MS - MLPA - Search for known variants: promotore MLH1, BRCA1, BRCA2 e RAD51C.	extraction + MS-MLPA	Recognized	PI 023 (ed.04,rev.1;2023)	0
F	Medical Genetics	FFPE	Search for unknown variants	--	Direct NGS Sequencing	Internal	--	0
D	Medical Genetics	FFPE	--	OncoPan® -Search for unknown somatic variants: ACD, APC, ATM, AXIN2, BAP1, BARD1, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDKN2A(α e β), CDK4(esone2), CHEK2, CTNNA1, EPCAM, FANCM, GREM1, HOXB13, MC1R, MITF(esone9), MLH1, MSH2, MSH3, MSH6, MUTYH, NBN, NTHL1, PALB2, PMS2, POLD1, POLE, POT1, PTEN, RAD51C, RAD51D, RNF43, SMAD4, STK11, TERF21P, TERT, TP53, KRAS, NRAS, BRAF, EGFR, HER2 (ERBB2), PIK3CA, c-KIT e PDGFR-α	extraction + NGS	Internal	PI 023 (ed.04,rev.1;2023)	0
D	Medical Genetics	FFPE	--	OncoHRD - Search for unknown variants: BRCA1, BRCA2 e HRD score	extraction + NGS	Internal	PI 023 (ed.04,rev.1;2023)	0
F	Medical Genetics	FFPE	Search for known variants	--	PCR + direct Sanger Sequencing	Internal	--	0
D	Medical Genetics	FFPE	--	Sanger Sequencing - Search for known variants: BRCA1, BRCA2, CDKN2A(alpha), MSH2, FANCM, POLE, RAD51D e TP53.	extraction + PCR + electrophoresis + sequencing	Internal	PI 023 (ed.04,rev.1;2023)	0
F	Medical Genetics	FFPE	Search for unknown variants	--	MLPA	Recognized	--	0
D	Medical Genetics	FFPE	--	MLPA - Search for unknown variants: TP53	extraction + MLPA	Recognized	PI 023 (ed.04,rev.1;2023)	0
F	Medical Genetics	FFPE	Search for known variants	--	MLPA	Recognized	--	0
D	Medical Genetics	FFPE	--	MLPA - Search for known variants: TP53, BRCA1 e BRCA2.	extraction + MLPA	Recognized	PI 023 (ed.04,rev.1;2023)	0
D	Medical Genetics	FFPE	--	MS - MLPA - Search for known variants: promotore MLH1, BRCA1, BRCA2 e RAD51C.	extraction + MS-MLPA	Recognized	PI 023 (ed.04,rev.1;2023)	0

Notes: Detail of examinations falling within the scope of UNI EN ISO 15189 accreditation issued by ACCREDIA, as per Annex rev0 to MED Accreditation Certificate No. 0015 M.

- 1) Letter F indicates the description of the flexible field. Letter D gives the list of detailed examinations.
- 2) Field filled only for detail examinations.
- 3) "Recognized" means method subject to review.
- 4) Code and revision of internal examination procedure is reported.
- 5) Category 0 means that the examination is performed in the laboratory.

Signature of the Laboratory Manager :

(Marco Alessandro Pierotti)