

F/D (1)	DISCIPLINE	SPECIMEN	TYPE OF EXAMINATION	DETAILED LIST (2)	METHOD /TECHNICS	TYPE OF PROCEDURE (3)	EXAMINATION PROCEDURE (4)	CATEGORY (5)
F	Medical Genetics	Whole blood	Search for known variants	--	PCR + direct Sanger Sequencing	B	--	0
D	Medical Genetics	Whole blood	--	<b>Sanger Sequencing - Search for known variants:</b> APC, ATM, BAP1, BARD1, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDKN2A(α e β), CDK4(esone2), CHEK2, CTNNNA1, DICER1, FANCM, FH, MC1R, MITF, MLH1, MSH2, MSH3, MSH6, MUTYH, NBN, NF1, NTHL1, PALB2, PIK3CA, PMS2, POLD1, POLE, PTEN, RAD51C, RAD51D, RET, SMAD4, STK1, VHL and TP53.	extraction + PCR + electrophoresis + sequencing	B	PI 026 rev.01	Veriti 96 DX (ThermoFisher), Sequencer 3500 DX (ThermoFisher)
F	Medical Genetics	Whole blood	Search for unknown variants	--	Direct NGS Sequencing	B	--	
D	Medical Genetics	Whole blood	--	<b>OncoPan® -Search for unknown germinal variants:</b> ACD, APC, ATM, AXIN2, BAP1, BARD1, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDKN2A(α e β), CDK4(esone2), CHEK2, CTNNNA1, 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	extraction + NGS	B	PI 023 rev.03	Biomek FX 1 (Beckman Coulter), NextSeq550DX (Illumina), MiSeqDX (Illumina)
D	Medical Genetics	Whole blood	--	<b>OncoPed - Search for unknown germinal variants:</b> APC, BAP1, BRCA1, BRCA2, CDH1, CDKN2A(α e β), CDK4(esone2), DICER1, EPCAM, FBXW7 FH, FLCN, MET, MITF, MLH1, MSH2, MSH3, MSH6, MUTYH, PALB2, PMS2, POT1, PRKAR1A, PTCH1, PTEN, RB1, SMARCA4, SMARCB1, SUFU, TP53, VHL and WT1.	extraction + NGS	B	PI 023 rev.03	Biomek FX 1 (Beckman Coulter), NextSeq550DX (Illumina), MiSeqDX (Illumina)
F	Medical Genetics	Whole blood	Search for unknown variants	--	MLPA	A	--	
D	Medical Genetics	Whole blood	--	<b>MLPA - Search for unknown germinal variants:</b> APC, ATM, BARD1, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDKN2A(α e β), CDK4, CHEK2, EPCAM, GREM1, MLH1, MSH2, MSH3, MSH6, MUTYH, PALB2, PMS2, PTEN, TP53, RAD50C, RAD51C, RAD51D, RB1, SMAD4, SMARCB1 and STK11.	extraction + MLPA	A	PI 027 rev.01	Veriti 96 DX (ThermoFisher), Sequencer 3500 DX (ThermoFisher)
F	Medical Genetics	Whole blood	Search for known variants	--	MLPA	A	--	
D	Medical Genetics	Whole blood	--	<b>MLPA - Search for known variants:</b> APC, ATM, BARD1, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDKN2A(α e β), CDK4, CHEK2, EPCAM, GREM1, MLH1, MSH2, MSH3, MSH6, MUTYH, PALB2, PMS2, PTEN, TP53, RAD50C, RAD51C, RAD51D, RB1, SMAD4, SMARCB1 and STK11.	extraction + MLPA	A	PI 027 rev.01	Veriti 96 DX (ThermoFisher), Sequencer 3500 DX (ThermoFisher)
D	Medical Genetics	Whole blood	--	<b>MS - MLPA - Search for known variants:</b> promotore MLH1, BRCA1, BRCA2 and RAD51C.	extraction + MS-MLPA	A	PI 027 rev.01	Veriti 96 DX (ThermoFisher), Sequencer 3500 DX (ThermoFisher)
F	Medical Genetics	FFPE	Search for unknown variants	--	Direct NGS Sequencing	B	--	
D	Medical Genetics	FFPE	--	<b>OncoPan® -Search for unknown somatic variants:</b> ACD, APC, ATM, AXIN2, BAP1, BARD1, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDKN2A(α e β), CDK4(esone2), CHEK2, CTNNNA1, 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 and PDGFRA.	extraction + NGS	B	PI 023 rev.03	Biomek FX 1 (Beckman Coulter), NextSeq550DX (Illumina), MiSeqDX (Illumina)
D	Medical Genetics	FFPE	--	<b>OncoHRD - Search for unknown variants:</b> BRCA1, BRCA2 and HRD score	extraction + NGS	B	PI 023 rev.03	Biomek FX 1 (Beckman Coulter), NextSeq550DX (Illumina)
F	Medical Genetics	FFPE	Search for known variants	--	PCR + direct Sanger Sequencing	B	--	
D	Medical Genetics	FFPE	--	<b>Sanger Sequencing - Search for known variants:</b> BRCA1, BRCA2, CDKN2A(alpha), MSH2, FANCM, POLE, RAD51D and TP53.	extraction + PCR + electrophoresis + sequencing	B	PI 026 rev.01	Veriti 96 DX (ThermoFisher), Sequencer 3500 DX (ThermoFisher)
F	Medical Genetics	FFPE	Search for unknown variants	--	MLPA	A	--	
D	Medical Genetics	FFPE	--	<b>MLPA - Search for unknown variants:</b> TP53	extraction + MLPA	A	PI 027 rev.01	Veriti 96 DX (ThermoFisher), Sequencer 3500 DX (ThermoFisher)
F	Medical Genetics	FFPE	Search for known variants	--	MLPA	A	--	
D	Medical Genetics	FFPE	--	<b>MLPA - Search for known variants:</b> TP53, BRCA1 and BRCA2.	extraction + MLPA	A	PI 027 rev.01	Veriti 96 DX (ThermoFisher), Sequencer 3500 DX (ThermoFisher)
D	Medical Genetics	FFPE	--	<b>MS - MLPA - Search for known variants:</b> promotore MLH1, BRCA1, BRCA2 and RAD51C.	extraction + MS-MLPA	A	PI 027 rev.01	Veriti 96 DX (ThermoFisher), Sequencer 3500 DX (ThermoFisher)

Note: Details of tests covered under the UNI EN ISO 15189 accreditation granted by ACCREDIA with accreditation certificate MED n. 0015 M.

1) The letter F indicates the description of the flexible scope. The letter D lists the detailed tests.

2) This field is completed only for detailed tests.

3) A refers to a method subject to verification, B refers to a method subject to validation by the laboratory.

4) The code and revision of the internal test procedure are provided.

5) Instrumentation

6) External quality assessment: name of VEQ/PT organizer, frequency of assessments/year

Signature of the Laboratory Manager :  
(Marco Alessandro Pierotti)

