



ردیف	Test	کلیدواژه keyword	ID	روش انجام method	نوع نمونه sample	حجم نمونه volume	شرایط نگهداری storage	زمان جوابدهی (روز) TAT (Day)	مدارک مورد نیاز document	نیاز به مشاوره قبل از انجام آزمایش دارد؟ Pre test counseling	نیاز به مشاوره بعد از انجام آزمایش دارد؟ Posttest counseling	قابلیت اورژانس دارد؟ Does it have an emergenc function
1	Achondroplasia	Diagnosis, FGFR3, Mutation Germline	Mol- Gene Sequencing	Sanger Sequencing	PBE or DNA	5-10 ml	2-8 °C	14	Radiography/ medical impression/ reports	Y	Y	N
2	ALK Common Somatic Mutation	Somatic Mutation, Neuroblastoma	Mol- Trans	Sanger Sequencing	PBE or DNA or paraffin block	5-10 ml/> 20% Tumoral Tissue	2-8 °C	14	Pathology report/ Patient clinical information/ Form TE-REX-FO-097	N	N	N
3	Alpha Thal Female	Alpha Globin, Carrier Detection, Hematologic diseases, Alpha Thalassemia	Mol- Thalasemia	Sanger Sequencing- Multiplex PCR	PBE or DNA	5-10 ml	2-8 °C	30	CBC- Hb Electrophoresis-Ferritin	Y	N	N
4	Alpha Thal Male	Alpha Globin, Carrier Detection, Hematologic diseases, Alpha Thalassemia	Mol- Thalasemia	Sanger Sequencing- Multiplex PCR	PBE or DNA	5-10 ml	2-8 °C	30	CBC- Hb Electrophoresis-Ferritin	Y	N	N
5	Alpha Thal Other	Alpha Globin, Carrier Detection, Hematologic diseases, Alpha Thalassemia	Mol- Thalasemia	Sanger Sequencing- Multiplex PCR	PBE or DNA	5-10 ml	2-8 °C	30	CBC- Hb Electrophoresis-Ferritin	Y	N	N
6	Alpha-Beta-XMN	Alpha Globin, Beta- Globin, Thalassemia, Hematologic diseases, Thalassemia Diagnosis, Thalassemia Major	Mol- Thalasemia	Sanger Sequencing- ARMS PCR- Multiplex PCR	PBE or DNA	5-10 ml	2-8 °C	30	CBC- Hb Electrophoresis-Ferritin- Medical impression- Transfusion dependency	Y	N	N



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7	ASXL1 Common Somatic Mutation	ASXL1, Somatic Mutation, Cancer, Myeloproliferative Neoplasms, (MPN)	Mol- Trans	Sanger Sequencing	PBE or DNA or paraffin block	5-10 ml/> 20% Tumoral Tissue	2-8 °C	14	medical impression/ reports/Pathology Reports/prescription	N	by the attending physician	N
8	ATRX Common Somatic Mutation	ATRX, Somatic Mutation, Nervous System Cancer	Mol- Trans	Sanger Sequencing	PBE or DNA or paraffin block	5-10 ml/> 20% Tumoral Tissue	2-8 °C	14	medical impression/ reports/Pathology Reports/prescription	N	by the attending physician	N
9	Beta Thal Female	Beta-Globin, Thalassemia; Hematologic diseases	Mol- Thalasemia	Sanger Sequencing- ARMS PCR	PBE or DNA	5-10 ml	2-8 °C	30	CBC- Hb Electrophoresis-Ferritin	Y	N	N
10	Beta Thal Male	Beta-Globin, Thalassemia; Hematologic diseases	Mol- Thalasemia	Sanger Sequencing- ARMS PCR	PBE or DNA	5-10 ml	2-8 °C	30	CBC- Hb Electrophoresis-Ferritin	Y	N	N
11	Beta Thal Other	Beta-Globin, Thalassemia; Hematologic diseases	Mol- Thalasemia	Sanger Sequencing- ARMS PCR	PBE or DNA	5-10 ml	2-8 °C	30	CBC- Hb Electrophoresis-Ferritin	Y	N	N
12	BRAF Mutation (Tissue)	BRAF, V600, Exon15, Somatic Mutation Nervous System Cancer, Melanoma, Colorectal Cancer, Lymphoma, Hairy cell leukemia	Mol- Trans	Sanger Sequencing	Tis or DNA or FFPE	5-10 ml/> 20% Tumoral Tissue	2-8 °C	7	medical impression/ reports/Pathology Reports/prescription	N	by the attending physician	N
13	CAH-Strip Assay	Diagnosis, big genitalia, CAH, Congenital adrenal hyperplasia	Mol- Others	Reverse Strip Assay	PBE or DNA	5-10 ml	2-8 °C	14	medical impression/ reports/Pathology Reports/prescription	Y	Y	N



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14	CALR	CALR, Somatic Mutation, Myeloproliferative Neoplasms (MPN), Cancer	Mol- Trans	Sanger Sequencing	PBE or DNA	5-10 ml	2-8 °C	14	Prescription, medical impression, CBC / reports	N	by the attending physician	N
15	CEBPA Common Somatic Mutation	CEBPA, Somatic Mutation, Acute Myeloid Leukemia (AML), Cancer	Mol- Trans	Sanger Sequencing	PBE or DNA or paraffin block	5-10 ml/> 20% Tumoral Tissue	2-8 °C	14	medical impression/ reports/Pathology Reports/prescription	N	by the attending physician	N
16	CF Common Mutation Female	CFTR gene (Cystic Fibrosis)	Mol- Others	Reverse Strip Assay	PBE or DNA	5-10 ml	2-8 °C	14	medical impression/ reports	Y	Y	N
18	CF Common Mutation Other	CFTR gene (Cystic Fibrosis)	Mol- Others	Reverse Strip Assay	PBE or DNA	5-10 ml	2-8 °C	14	medical impression/ reports	Y	Y	N
19	CKIT	CKIT, Somatic Mutation, Acute Myeloid Leukemia (AML), Myeloproliferative Neoplasms (MPN), Sarcoma, Melanoma, Cancer	Mol- Trans	Sanger Sequencing	PBE or DNA	5-10 ml	2-8 °C	14	medical impression/ reports/Pathology Reports/prescription	N	by the attending physician	N
20	CKIT-Tissue	CKIT, Somatic Mutation, Gastric Cancer, GIST , Cancer	Mol- Trans	Sanger Sequencing	Tis or DNA or FFPE	> 20% Tumoral Tissue	2-8 °C	14	medical impression/ reports/Pathology Reports/prescription	N	by the attending physician	N



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21	Clotting Panel	MTHFR (A1298C) (C677T), PAI, FV;FXIII(V34L in Factor XIII gene), Prothrombin (Factor II) (G20210A),	Mol- Others	Reverse Strip Assay	PBE or DNA	5-10 ml	2-8 °C	14	medical impression/ reports	N	by the attending physician	N
22	Connexin 26 Female	GJB2- Connexin26- 35Del G, Diagnosis, Carrier Detection	Mol- Gene Sequencing	Sanger Sequencing	PBE or DNA	5-10 ml	2-8 °C	30	Audiometry result	Y	Y	N
23	Connexin 26 Male	GJB2- Connexin26- 35Del G, Diagnosis, Carrier Detection	Mol- Gene Sequencing	Sanger Sequencing	PBE or DNA	5-10 ml	2-8 °C	30	Audiometry result	Y	Y	N
24	Connexin 26 Other	GJB2- Connexin26- 35Del G, Diagnosis, Carrier Detection	Mol- Gene Sequencing	Sanger Sequencing	PBE or DNA	5-10 ml	2-8 °C	30	Audiometry result	Y	Y	N
25	Connexin 30	GJB6- Connexin30, Diagnosis, Carrier	Mol- Gene Sequencing	Sanger Sequencing	PBE or DNA	5-10 ml	2-8 °C	30	Audiometry result	Y	Y	N
26	CYP2C9 Genotype	MAYZENT, Multiple Sclerosis, MS	Mol- Trans	Sanger Sequencing	PBE or DNA	5-10 ml	2-8 °C	14	medical impression/ reports/Pathology Reports/prescription	Y	N	N
27	DM1	DM1	Mol- Others	Fragment Analysis	PBE or DNA	5-10 ml	2-8 °C	30	medical impression/ reports	Y	Y	N
28	DNA Bank	DNA Bank	Mol- Extraction	DNA Extraction	PBE or Tis	5-10 ml Blood or 20% Tumoral Tissue	2-8 °C	1095	Prescription	Y	Y	N
29	DNA Collection- mouth swab		Mol- Extraction	DNA Extraction			2-8 °C			Y	N	N



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30	DNA-Extraction- Report	Checking of Quality and Quantity of DNA	Mol- Extraction	DNA Extraction	PBE or Tis	5-10 ml Blood or 20% Tumoral Tissue	2-8 °C	3	medical impression/ reports	Y	N	N
31	DNMT3A Common Somatic Mutation	DNMT3A, Common Somatic Mutation, Pharmacogenetics, Myeloproliferative Neoplasms (MPN), Acute Myeloi Leukemia (AML)	Mol- Trans	Sanger Sequencing	PBE or Tis or DNA or FFPE	5-10 ml/> 20% Tumoral Tissue	2-8 °C	14	medical impression/ reports/Pathology Reports/prescription	N	by the attending physician	N
32	DPYD (5-fu)	colorectal cancer , Solid tumor , IVS14+1G>A	Mol- Others	Reverse Strip Assay	PBE or DNA	5-10 ml	2-8 °C	14	medical impression/ reports	N	by the attending physician	N
33	Duchenne-Ind	Duchenne Muscular Dystrophy, DMD, BMD, Dystrophin. Deletion /Duplication, Diagnosis, Carrier Detection	Mol- Others	MLPA	PBE or DNA	5-10 ml	2-8 °C	14	medical impression/ reports	Y	Y	N
34	EGFR	Lung Cancer, Non Small Cell Carcinoma, Somatic Mutation, Exon18, 19, 20, 21	Mol- Trans	Sanger Sequencing/ Reverse Strip Assay	Tis or DNA or FFPE	> 20% Tumoral Tissue	2-8 °C	7	medical impression/ reports/Pathology Reports/prescription	N	N	N
35	ESR1	Breast cancer, Prostat cancer, Estrogen receptor	Mol- Trans	Sanger Sequencing	Tis or FFPE	> 20% Tumoral Tissue	2-8 °C	14	medical impression/ reports/Pathology Reports/prescription	N	N	N



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36	Factor XIII	FXIII(V34L in Factor XIII gene, Diagnosis, Carrier Detection	Mol- Others	Sanger Sequencing/ Reverse Strip Assay	PBE or DNA	5-10 ml	2-8 °C	14	medical impression/ reports	N	N	N
37	Factor-V (Leiden)	Factor-V (Leiden) - G1691A	Mol- Others	Sanger Sequencing/ Reverse Strip Assay	PBE or DNA	5-10 ml	2-8 °C	14	medical impression/ reports	N	N	N
38	FancA-Seq- MLPA	FancA. Fanconi anemi, Germline Mutation	Mol- Gene Sequencing	Sanger Sequencing/ML PA	PBE or DNA	5-10 ml	2-8 °C	60	medical impression/ reports	Y	Y	N
39	FAP (APC gene mutation analysis)	Cancer, Pericardial, Peritoneal, Screening, Slide, Urine	Mol- Gene Sequencing	Sanger Sequencing	PBE or DNA	5-10 ml	2-8 °C	60	-	Y	by the attending physician	N
40	FLT-3	FLT3-Acute MyeloiLeukemia (AML), FLT3-ITD, FLT3-835-Somatic Mutation	Mol- Trans	Sanger Sequencing	PBE or DNA	5-10 ml	2-8 °C	14	CBC/medical impression/ reports/Pathology Reports/prescription	N	by the attending physician	N
41	FMF	FMF-Familial Mediterranean Fever, MeFv Mutation, Common Mutation, Diagnosis, Carrier Detection	Mol- Others	Reverse Strip Assay	PBE or DNA	5-10 ml	2-8 °C	14	medical impression/ reports	N	Y	N
42	Fragment Reaction	Diagnostic test/ Research	Mol- Sequencing	Fragment Analysis	PCR Product	25UL	2-8 °C	3	لیبل نمونه روی میکروتیوپ و اطلاعات فرگمنت در اکسل ارسالی	N	N	N



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43	Fragment Run	Diagnostic test/ Research	Mol- Sequencing	Fragment Analysis	PCR Product	25UL	2-8 °C	3	لیبل نمونه روی میکروتیوپ و اطلاعات فرگمنت در اکسل ارسالی	N	N	N
44	FraX-Mol	FraX, Premature Ovarian Failure (POF), X link Syndromic Mental Retardation, Diadnosis, Carrier Detection	Mol- Others	PCR- Fragment Analysis/Southern	PBE or DNA	5-10 ml	2-8 °C	30	medical impression/ reports	Y	Y	N
45	Friedreich`s ataxia	FXN, GAA Expansion, Friedreich`s ataxia	Mol- Others	PCR- Fragment Analysis	PBE or DNA	5-10 ml	2-8 °C	30	medical impression/ reports	Y	Y	N
46	GATA Common Somatic Mutation	Common Somatic Variant, Myeloproliferative	Mol- Trans	Sanger Sequencing	PBE or DNA	5-10 ml	2-8 °C	14	medical impression/ reports/Pathology Reports/prescription	N	Y	N
47	Gaucher	GBA gene;beta- glucocerebrosidase, Dagnosis, Carrier Detection	Mol- Others	Reverse Strip Assay	PBE or DNA	5-10 ml	2-8 °C	14	medical impression/ reports	N	Y	N
48	Gene Seq 02-05 Exon	Gemline Mutation, Diagnosis, Carrier Detection	Mol- Gene Sequencing	Sanger Sequencing	PBE or DNA	5-10 ml	2-8 °C	30	Prescription, medical impression/ reports	Y	Y	N
49	Gene Seq 06-10 Exon	Gemline Mutation, Diagnosis, Carrier Detection	Mol- Gene Sequencing	Sanger Sequencing	PBE or DNA	5-10 ml	2-8 °C	30	medical impression/ reports	Y	Y	N



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50	Gene Seq 11-25 Exon	Gemline Mutation, Diagnosis, Carrier Detection	Mol- Gene Sequencing	Sanger Sequencing	PBE or DNA	5-10 ml	2-8 °C	60	medical impression/ reports	Y	Y	N
51	Gene Seq 26-50 Exon	Gemline Mutation, Diagnosis, Carrier Detection	Mol- Gene Sequencing	Sanger Sequencing	PBE or DNA	5-10 ml	2-8 °C	60	medical impression/ reports	Y	Y	N
52	H3F3A Common Somatic Mutation	H3F3A, Common Somatic Mutation, Pharmacogenetics, Nervous System Cancer, Cancer	Mol- Trans	Sanger Sequencing	Tis or DNA or FFPE	> 20% Tumoral Tissue	2-8 °C	14	medical impression/ reports/Pathology Reports/prescription	N	by the attending physician	N
53	Hemochromatosis	HFE, Diagnosis, Carrier Detection	Mol- Others	Reverse Strip Assay	PBE or DNA	5-10 ml	2-8 °C	14	medical impression/ reports	Y	Y	N
54	Hemophilia A- Inversion 22	Inversion 22	Mol- Gene Sequencing	Long Range PCR	PBE or DNA	5-10 ml	2-8 °C	30	medical impression/ reports/Pathology Reports/prescription	Y	Y	N
55	Hemophilia A- Sequencing	Sequencing	Mol- Gene Sequencing	Sanger Sequencing	PBE or DNA	5-10 ml	2-8 °C	60	medical impression/ reports/Pathology Reports/prescription	Y	Y	N
56	Huntington	HTT gene, CAG Expanation, Huntington's disease (HD), autosomal dominant inheritanc	Mol- Others	PCR- Fragment Analysis	PBE or DNA	5-10 ml	2-8 °C	30	medical impression/ reports	Y	Y	N
57	Hypochondroplasia	FGFR3, Germline Mutation, Diagnosis, Carrier Detection	Mol- Gene Sequencing	Sanger Sequencing	PBE or DNA	5-10 ml	2-8 °C	14	medical impression/ reports	Y	Y	N





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58	IDH1 Common Somatic Mutation	IDH1, Somatic Mutation, Nervous system Cancer, Myeloproliferative Neoplasms (MPN)	Mol- Trans	Sanger Sequencing	Tis or DNA or FFPE	> 20% Tumoral Tissue	2-8 °C	7	medical impression/ reports/Pathology Reports/prescription	N	by the attending physician	N
59	IDH2 Common Somatic Mutation	IDH2, Somatic Mutation, Nervous system Cancer, Myeloproliferative Neoplasms (MPN)	Mol- Trans	Sanger Sequencing	Tis or DNA or FFPE	> 20% Tumoral Tissue	2-8 °C	7	medical impression/ reports/Pathology Reports/prescription	N	by the attending physician	N
60	Imatinib Resistance	CML, T315I, resistance to tyrosine kinase inhibitor (TKI) therapy,	Mol- Trans	Sanger Sequencing	PBE or cDNA	5-10 ml	2-8 °C	14	CBC/ Prescription/ medical impression/ reports	N	by the attending physician	N
61	Irinotecan Toxicity	UGT1A1*28 genotype, Pharmacogenomics, Pancreatic Neoplasms, Colorectal Neoplasms,	Mol- Others	Sanger Sequencing	PBE or DNA	5-10 ml	2-8 °C	14	medical impression/ reports	N	جواب غیر طبیعی	N
62	JAK2 Exon 12	JAK2, Myeloproliferative Neoplasms (MPN), Leukemia, MPN	Mol- Trans	Sanger Sequencing	PBE or DNA	5-10 ml	2-8 °C	7	CBC/ Prescription/ medical impression/ reports	N	by the attending physician	N
63	JAK2 gene v617F Mutation	JAK2, V617F, Leukemia, Myeloproliferative Neoplasms (MPN), CML, AML	Mol- Trans	Sanger Sequencing/ Real time PCR	PBE or DNA	5-10 ml	2-8 °C	14	CBC/ Prescription/ medical impression/ reports	N	by the attending physician	N



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64	K-RAS	KRAS Common Mutation, Exon 2, 3, 4, Codone 12-13.61-colorectal Cancer	Mol- Trans	Sanger Sequencing/ Reverse Strip Assay/Real time PCR	Tis Or DNA or FFPE	> 20% Tumoral Tissue	2-8 °C	7-10	medical impression/ reports/Pathology Reports/prescription	N	by the attending physician	N
65	MCC+QF-PCR Aneuploidies	Maternal Cell Contamination	Mol- Trans	Fragment Analysis	AF/AFC/Tis/ PBE or DNA	5-10 ml	2-8 °C	3	Prescription/ medical impression/ reports	Y	Y	N
66	MCC-PND	Maternal Cell Contamination	Mol- Trans	Fragment Analysis	PBE or DNA	5-10 ml	2-8 °C	3	Prescription/ medical impression/ reports	Y	Y	N
67	MLPA- CAPN3	MLPA- Large Deletion /Insertion, Diagnosis, Carrier Detection, CAPN3, CALpain3	Mol- Others	MLPA	PBE or DNA	5-10 ml	2-8 °C	14	Prescription/ medical impression/ reports	Y	Y	N
68	MLPA- Celiac DQ2 & DQ8	Celiac disease (HLA-DQ2/ HLA-DQ8)	Mol- Others	MLPA	PBE or DNA	5-10 ml	2-8 °C	14	Prescription/ medical impression/ reports	Y	N	N
69	MLPA-Alpha	MLPA, Alpha Thalassemia, Detection of copy number changes in the Alpha globin gene cluster	Mol- Thalasemia	MLPA	PBE or DNA or CV or AF or AFC	5-10 ml	2-8 °C	14	CBC- Hb Electrophoresis	Y	Y	N
70	MLPA- Aneuploidies	MLPA-Aneuploidies, QF, Down Syndrome	Mol- Others	MLPA	PBE or DNA or CV or AF or AFC	5-10 ml	2-8 °C	7	Prescription/ medical impression/ reports	Y	Y	N



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71	MLPA-Beta	MLPA, Delta-Beta Thalassemia, Detection of copy number changes in the Beta globin gene cluster	Mol- Thalasemia	MLPA	PBE or DNA or CV or AF or AFC	5-10 ml	2-8 °C	14	CBC- Hb Electrophoresis	Y	Y	N
72	MLPA-BRCA I-II	MLPA, Breast and Ovarian Cancer, Diagnosis, Carrier Detection	Mol- Others	MLPA	PBE or DNA	5-10 ml	2-8 °C	14	medical impression/ reports	Y	Y	N
73	MLPA-CLL	deletion, Insertion, Chronic lymphocytic leukemia	Mol- Trans	MLPA	PBE or DNA	5-10 ml	2-8 °C	14	Prescription/ medical impression/ reports	Y	Y	N
74	MLPA-CMT (Duplication- PMP22)	MLPA, Charcot-Marie-Tooth disease (CMT), HNPP, Diagnosis, Carrier Detection	Mol- Others	MLPA	PBE or DNA	5-10 ml	2-8 °C	14	medical impression/ reports	Y	Y	N
75	MLPA-Del/Dup I	MLPA- Large Deletion /Insertion, Diagnosis, Carrier Detection	Mol- Others	MLPA	PBE or DNA	5-10 ml	2-8 °C	120	medical impression/ reports	Y	Y	N
76	MLPA-MGMT Methylation	Hypermethylation, glioma	Mol- Trans	MLPA	Tis Or DNA or FFPE	> 20% Tumoral Tissue/5-10 ml	2-8 °C	14	medical impression/ reports/Pathology Reports/prescription	N	by the attending physician	Y



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77	MLPA- Microdeletions	MLPA- Deletion /Insertion, Diagnosis, Carrier Detection 1p36 deletion syndrome/2p16.1-p15 microdeletion syndrome/2q23.1 microdeletion/microdupli cation syndrome/Glass syndrome/3q29 microdeletion syndrome/3q29 microduplication syndrome/Wolf- Hirschhorn syndrome/Cri- du-Chat syndrome/Sotos syndrome/Williams- Beuren syndrome/Williams- Beuren duplication syndrome/Langer- Giedion syndrome/9q22.3 microdeletion syndrome/DiGeorge syndrome-2/Prader-Willi syndrome/Angelman syndrome/Witteveen- Kolk* / 15q24 microdeletion syndrome/Rubinstein-	Mol- Others	MLPA	PBE or DNA	5-10 ml	2-8 °C	14	medical impression/ reports	N	N	N



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78	MLPA-MLH1- 2,PMS2,MSH6 Hypermethylation	Hypermethylation, Somatic Mutation, Mis match repair gene, microsatellite instability	Mol- Trans	MLPA	Tis Or DNA or FFPE	> 20% Tumoral Tissue/5-10 ml	2-8 °C	14	medical impression/ reports/Pathology Reports/prescription	Y	by the attending physician	N
79	MLPA-Mutation CAH	Diagnosis, big genitalia, CAH, Congenital adrenal hyperplasia	Mol- Others	MLPA	PBE or DNA	5-10 ml	2-8 °C	14	medical impression/ reports/Pathology Reports/prescription	N	by the attending physician	N
80	MLPA-SGCA	Limb-girdle muscular dystrophy; MLPA- Large Deletion Duplication, Diagnosis, Carrier Detection	Mol- Others	MLPA	PBE or DNA	5-10 ml	2-8 °C	14	medical impression/ reports	Y	Y	N
81	MPL-Mutation	MPL, Somatic Mutation- Myeloproliferative Neoplasms (MPN), Cancer	Mol- Trans	Sanger Sequencing	PBE or DNA	5-10 ml	2-8 °C	14	CBC/ Prescription/ medical impression/ reports	N	by the attending physician	N
82	MSI	Micro Satellite Inastability- Colorectal Cancer , MMR, Miss Match Repair, HNPCC	Mol- Trans	Fragment Analysis	PBE and Tis or FFPE	5-10 ml/> 20% Tumoral Tissue	2-8 °C	14	medical impression/ reports/Pathology Reports/prescription	N	by the attending physician	N
83	MTH-FR	MTHFR (A1298C) (C677T)	Mol- Others	Sanger Sequencing/ Reverse Strip Assay	PBE or DNA	5-10 ml	2-8 °C	14	medical impression/ reports	N	by the attending physician	N



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84	Mutation Confirmation-Family (up to 3 person)	Mutation Confirmation, Diagnosis, Carrier Detection	Mol- Gene Sequencing	Sanger Sequencing	PBE or DNA	5-10 ml	2-8 °C	60	Prescription/ medical impression/ reports	Y	Y	N
85	Mutation Confirmation-Ind (additional person)	Mutation Confirmation, Diagnosis, Carrier Detection	Mol- Gene Sequencing	Sanger Sequencing	PBE or DNA	5-10 ml	2-8 °C	60	Prescription/ medical impression/ reports	Y	Y	N
86	NF1 Common Somatic Mutation	NF1, Somatic Mutation, Sarcoma, Cancer	Mol- Trans	Sanger Sequencing	Tis Or DNA or FFPE	> 20% Tumoral Tissue	2-8 °C	14	medical impression/ reports/Pathology Reports/prescription	N	by the attending physician	N
87	NGS- Carrier Screening-Medical Exome	Carrier Screening, OMIM Genes	Mol- NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	90-120	Prescription/ medical impression/ reports/pedigree	Y	Y	N
88	NGS- Multi Cancer With CNV Panel (Solid Tumor and Non solid Tumor Cancer)	Solid Tumor, Cancer, NGS, Germline, Somatic	Mol- NGS	NGS	PBE or DNA or Tis or FFPE	5-10 ml/> 20% Tumoral Tissue	2-8 °C	90-120	Prescription/ medical impression/ reports	Y	Y	N
89	NGS-Autism Spectrum Disorders-Exome	Autism, ASD, intellectual disability	Mol- NGS	NGS	PBE or DNA or Tis or FFPE	5-10 ml/> 20% Tumoral Tissue	2-8 °C	120	Prescription/ medical impression/ reports	Y	Y	N
90	NGS-Ataxia Exome	NGS, Ataxia, Germline Mutation, Diagnosis, Carrier Detection	Mol- NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	90-120	Prescription/ medical impression/ reports	Y	Y	N



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91	NGS-BRCA I-II Panel	NGS- Germline mutation, Diagnosis, Breast cancer, Ovarian Cancer, pancreatic cancer, BRCA1- BRCAII, BRCA1, BRCA2, Sequencing	Mol- Gene Sequencing	NGS	PBE or DNA	5-10 ml	2-8 °C	60	Prescription/ medical impression/ reports	Y	Y	N
92	NGS-BRCA I-II Plus del/dup	NGS- Germline mutation, Diagnosis, Breast cancer, Ovarian Cancer, pancreatic cancer, BRCA1- BRCAII, BRCA1, BRCA2, Sequencing	Mol- Gene Sequencing	NGS	PBE or DNA	5-10 ml	2-8 °C	60	Prescription/ medical impression/ reports	Y	Y	N
93	NGS-BRCA I-II Plus del/dup (Somatic) Panel	NGS- Germline mutation, Diagnosis, Breast cancer, Ovarian Cancer, pancreatic cancer, BRCA1- BRCAII, BRCA1, BRCA2, Sequencing	Mol- Trans	NGS	DNA in streck tubes	5-10 ml	2-8 °C	60	Prescription/ medical impression/ reports	Y	Y	N
94	NGS-Carrier Screening Female	NGS,Parents (Including carrier detection), Gemline Mutation, Diagnosis, Carrier Detection, Reproductive Screening	Mol- NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	90-120	Prescription/ medical impression/ reports	Y	Y	N



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95	NGS-Carrier Screening Female (Without DMD)	NGS,Parents (Including carrier detection), Gemline Mutation, Diagnosis, Carrier Detection, Reproductive Screening	Mol- NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	90-120	Prescription/ medical impression/ reports	Y	Y	N
96	NGS-Carrier Screening male	NGS,Parents (Including carrier detection), Gemline Mutation, Diagnosis, Carrier Detection, Reproductive Screening	Mol- NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	90-120	Prescription/ medical impression/ reports	Y	Y	N
97	NGS- Ciliopathies Exome	NGS-Ciliopathies, Gemline Mutation, Diagnosis, Carrier Detection	Mol- NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	90-120	Prescription/ medical impression/ reports	Y	Y	N
98	NGS-CMT & Sensory Neuropathies (103G) Exome	NGS, Sensory Neuropathies, Gemline Mutation, Diagnosis, Carrier Detection	Mol- NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	90-120	Prescription/ medical impression/ reports	Y	Y	N
99	NGS-CNV Exome	NGS, Gemline Mutation, Diagnosis, Carrier Detection, Solid Tumor, Cancer	Mol- NGS	NGS	PBE or DNA or Tis or FFPE	5-10 ml/> 20% Tumoral Tissue	2-8 °C	120	Prescription/ medical impression/ reports	Y	Y	N





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100	NGS-Corpus Callosum Agenesis Exome	NGS-Corpus Callosum Agenesis, Gemline Mutation, Diagnosis, Carrier Detection	Mol- NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	90-120	Prescription/ medical impression/ reports	Y	Y	N
101	NGS-Craniofacial anomalies Exome	NGS, Carniofacial Anomalies, Gemline Mutation, Diagnosis, Carrier Detection	Mol- NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	90-120	Prescription/ medical impression/ reports	Y	Y	N
102	NGS-Disorders in sex development Exome	NGS, Disorders in sex development, Gemline Mutation, Diagnosis, Carrier Detection	Mol- NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	90-120	Prescription/ medical impression/ reports	Y	Y	N
103	NGS-Duo (Proband & 1 family member) Exome	NGS, Duo (Proband & 1 family member, Gemline Mutation, Diagnosis, Carrier Detection	Mol- NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	90-120	Prescription/ medical impression/ reports	Y	Y	N
104	NGS-Epilepsy Exome	NGS-Epilepsy, Gemline Mutation, Diagnosis, Carrier Detection	Mol- NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	90-120	Prescription/ medical impression/ reports	Y	Y	N
105	NGS-Exome (Medical exome for OMIM referenced genes 200X)	NGS, Gemline Mutation, Diagnosis, Carrier Detection	Mol- NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	90-120	Prescription/ medical impression/ reports	Y	Y	N



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106	NGS-Exome (Medical exome for OMIM referenced genes)	NGS-Exome (Medical exome for OMIM referenced genes), Gemline Mutation, Diagnosis, Carrier Detection	Mol- NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	90-120	Prescription/ medical impression/ reports	Y	Y	N
107	NGS-Exome+ Mito 100X	Mitochondrial disorder, hetroplasmly, mt DNA Screening	Mol- NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	90-120	Prescription/ medical impression/ reports	Y	Y	N
108	NGS-Hearing Loss Exome	NGS-Hearing Loss, Gemline Mutation, Diagnosis, Carrier Detection	Mol- NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	90-120	Prescription/ medical impression/ reports	Y	Y	N
109	NGS- Hemostatic/Thro mbotic disorders Exome	NGS- Hemostatic/Thrombotic disorders, Gemline Mutation, Diagnosis, Carrier Detection	Mol- NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	90-120	Prescription/ medical impression/ reports	Y	Y	N
110	NGS-Hereditary Cancer	NGS,Hereditary Cancer, Gemline Mutation, Diagnosis, Carrier Detection	Mol- NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	90-120	Prescription/ medical impression/ reports	Y	Y	N
111	NGS-Hereditary comprehensive cancer panel	NGS,Hereditary Cancer,	Mol-Gene seq	NGS	PBE or DNA	5-10 ml	2-8 °C	60	Prescription/ medical impression/ reports	Y	Y	N



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112	NGS-Hereditary Cancer plus CNV	NGS,Hereditary Cancer, Gemline Mutation, Diagnosis, Carrier Detection,CNV	Mol- NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	60	Prescription/ medical impression/ reports	Y	Y	N
113	NGS-Hereditary Spastic Paraplegia Exome	NGS, Hereditary Spastic Paraplegia Gemline Mutation, Diagnosis, Carrier Detection	Mol- NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	90-120	Prescription/ medical impression/ reports	Y	Y	N
114	NGS-ID (Syndromic- NonSyndromic) Exome	NGS-ID (Syndromic- NonSyndromic), Gemline Mutation, Diagnosis, Carrier Detection	Mol- NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	90-120	Prescription/ medical impression/ reports	Y	Y	N
115	NGS-IBMFS (Aplastic anemia)	Aplastic anemia, Fanconi anemia, shmachman diamond syndrome, diamond blackfan syndrome, bone marrow failure	Mol- NGS	NGS	PBE or DNA or Tis or FFPE	5-10 ml/> 20% Tumoral Tissue	2-8 °C	60	Prescription/ medical impression/ reports	Y	Y	N
116	NGS-Iron disorders Exome	NGS,Iron disorders, Gemline Mutation, Diagnosis, Carrier Detection	Mol- NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	90-120	Prescription/ medical impression/ reports	Y	Y	N



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117	NGS-Mendelian Disorders Exome	NGS, Mendelian Disorders, Gemline Mutation, Diagnosis, Carrier Detection	Mol- NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	90-120	Prescription/ medical impression/ reports	Y	Y	N
118	NGS-Metabolic disorders Exome	NGS-Metabolic disorders, Metabolic disorders	Mol- NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	90-120	Prescription/ medical impression/ reports	Y	Y	N
119	NGS-Mitochondrial disorders Exome	NGS, Mitochondrial disorders, Gemline Mutation, Diagnosis, Carrier Detection	Mol- NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	90-120	Prescription/ medical impression/ reports	Y	Y	N
120	NGS-Movement disorders Exome	NGS, Movement disorders, Gemline Mutation, Diagnosis, Carrier Detection	Mol- NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	90-120	Prescription/ medical impression/ reports	Y	Y	N
121	NGS-Multiple congenital anomalies Exome	NGS, Multiple congenital anomalies, Gemline Mutation, Diagnosis, Carrier Detection	Mol- NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	90-120	Prescription/ medical impression/ reports	Y	Y	N
122	NGS-Muscular Dystrophy & Muscledisorders Exome	NGS, Muscular Dystrophy & Muscledisorders, Gemline Mutation, Diagnosis, Carrier Detection	Mol- NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	90-120	Prescription/ medical impression/ reports	Y	Y	N



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123	NGS-Onco Myeloid Panel	KDM6A, Leukemia, NGS, Cancer, TP53, SMC1A, PTEN, NMP1, HRAS, ETV6, CEBPA, BRAF, FLT3, JAK2, U2AF1, WT1, SF3B1, ATRX, ASXL1, K1T, NRAS, E2H2, IDH1, IDH2	Mol- NGS	NGS	PBE or DNA	10 ml	2-8 °C	30	Prescription/ medical impression/ reports	Y	Y	N
124	NGS-Onco Multi cancer RNA fusion panel	Leukemia, RNA, NGS, Cancer, RAF1, BRAF, NTRK1, 2, 3, EGFR, FGFR3, PBX1, NRG, ROS1, RET, ALK, TFE3, PRKACA, ERG, PPARG, MET	Mol- NGS	NGS	PBE or FFPE	10 ml	2-8 °C	30	Prescription/ medical impression/ Pathology reports	Y	Y	N
125	NGS-Parents (Including carrier detection) Exome	NGS, Parents (Including carrier detection), Gemline Mutation, Diagnosis, Carrier Detection	Mol- NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	90-120	Prescription/ medical impression/ reports	Y	Y	N
126	NGS-Primary immunodeficiencies Exome	NGS-Primary immunodeficiencies, Gemline Mutation, Diagnosis, Carrier Detection	Mol- NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	90-120	Prescription/ medical impression/ reports	Y	Y	N
127	NGS-Reanalysis of data	NGS, Reanalysis of data, Gemline Mutation, Diagnosis, Carrier Detection	Mol- NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	60	Prescription/ medical impression/ reports	Y	Y	N



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128	NGS-Research Exome 100X	NGS,Research, Gemline Mutation, Diagnosis, Carrier Detection	Mol- NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	90-120	Prescription/ medical impression/ reports	Y	Y	N
129	NGS-RP Exome	NGS,Gemline Mutation, Diagnosis, Carrier Detection	Mol- NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	90-120	Prescription/ medical impression/ reports	Y	Y	N
130	NGS-Skin disorders Exome	NGS,Skin Disorders, Gemline Mutation, Diagnosis, Carrier Detection	Mol- NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	90-120	Prescription/ medical impression/ reports	Y	Y	N
131	NGS-TMB and MSI	Thyroid cancer, Lung, Colorectal cancer, EGFR Mutation	Mol- NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	45	Prescription/ medical impression/ reports	Y	Y	N
132	NGS-Trio (Proband & 2 family preferred members) Exome	NGS-Trio (Proband & 2 family preferred members) Exome, Gemline Mutation, Diagnosis, Carrier Detection	Mol- NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	90-120	Prescription/ medical impression/ reports	Y	Y	N
133	NGS-Vision disorders Exome	NGS,Gemline Mutation, Diagnosis, Carrier Detection, Vision disorders	Mol- NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	90-120	Prescription/ medical impression/ reports	Y	Y	N
134	NGS-Whole Genome Sequencing (WGS)	Whole Genome Sequencing, Breakpoint detection	Mol- NGS	Whole genome sequencing	PBE or DNA	5-10 ml	2-8 °C	90	medical impression/ reports	Y	Y	N



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135	NPM-1	NPM1, Somatic Mutation	Mol- Trans	Sanger Sequencing	PBE or DNA	5-10 ml	2-8 °C	7	Prescription/medical impression/ reports/CBC	N	by the attending physician	N
136	N-RAS	NRAS Common Mutation, Exon 2, 3, 4, Codone 12-13.61- colorectal Cancer	Mol- Trans	Sanger Sequencing/ Reverse Strip Assay/Real time PCR	Tis Or DNA	> 20% Tumoral Tissue	2-8 °C	7-10	medical impression/ reports/Pathology Reports/prescription	N	by the attending physician	N
137	PAI	PAI, Clotting Problem, Trombosis	Mol- Others	Sanger Sequencing/ Reverse Strip Assay	PBE or DNA	5-10 ml	2-8 °C	14	medical impression/ reports	N	by the attending physician	N
138	Panel ALL	t(1;19) - t(12, 21) - t(9;22); t(4;11), Translocation , Leukemia, Diagnosis, Prognosis	Mol- Trans	RT-PCR/Real Time PCR	PBE or BM	5-10 ml	2-8 °C	14	All disease diagnosis documents - prescription - CBC	N	by the attending physician	N
139	Panel AML	t(4;11) - t(8, 21) - t(9;22); t(15;71), inv16, Translocation , Leukemia, Diagnosis, Prognosis	Mol- Trans	RT-PCR/Real Time PCR	PBE or BM	5-10 ml	2-8 °C	14	All disease diagnosis documents - prescription - CBC	N	by the attending physician	N
140	Paternity	Paternity	Mol- Trans	Fragment Analysis	PBE or DNA	5-10 ml	2-8 °C	14	Identity documents	Y	Y	N
141	Paternity-Ind	Paternity	Mol- Trans	Fragment Analysis	PBE or DNA	5-10 ml	2-8 °C	14	Identity documents	Y	Y	N
142	PCR- Ampilification	PCR	Mol- Trans	PCR	DNA in streck tubes	5-10 ml	2-8 °C	3	Prescription/ medical impression/ reports	N	Y	N



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143	PDGFRa-Tissue	PDGFRa, Somatic Mutation, GIST (Exon18), Sarcoma	Mol- Trans	Sanger Sequencing	Tis Or DNA	> 20% Tumoral Tissue	2-8 °C	14	medical impression/ reports/Pathology Reports/prescription	N	by the attending physician	N
144	PGD	PGD-PGT-M-Monogenic disease-IVF-PND	Mol- PGD	Sanger sequencing/ Linkage analysis	Blastomere/ blastocyst	1-4 Blastomeres	2-8 °C	2	medical impression/ reports	Y	Y	Y
145	PGD I	PGD-PGT-M-Monogenic disease-IVF-PND	Mol- PGD	Sanger sequencing/ Linkage analysis	Blastomere/ blastocyst	<4 Blastomeres	2-8 °C	2	medical impression/ reports	Y	Y	*Y
146	PGD II	PGD-PGT-M-Monogenic disease-IVF-PND	Mol- PGD	Sanger sequencing/ Linkage analysis	Blastomere/ blastocyst	>4 Blastomeres	2-8 °C	2	medical impression/ reports	Y	Y	N
147	PGD SEX I	PGD-PGT-M-Monogenic disease-IVF-PND-Pre-PGT-M-SEX determination	Mol- PGD	AMX/SRY amplification-QF analysis	Blastomere/ blastocyst	1-4 Blastomeres	2-8 °C	2	medical impression/ reports	Y	Y	Y
148	PGD SEX II	PGD-PGT-M-Monogenic disease-IVF-PND-Pre-PGT-M-SEX determination	Mol- PGD	AMX/SRY amplification-QF analysis	Blastomere/ blastocyst	>4 Blastomeres	2-8 °C	2	medical impression/ reports	Y	Y	Y





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149	PGD-Pre	PGD-PGT-M-Monogenic disease-IVF-PND-Pre- PGT-M	Mol- PGD	Sanger sequencing/ Linkage analysis	PBE or DNA	5-10 ml	2-8 °C	60	Prescription/ medical impression/ reports	Y	Y	N
150	PGD-Pre Beta & GJB2	PGD-PGT-M-Monogenic disease-IVF-PND-Pre- PGT-M	Mol- PGD	Sanger sequencing/ Linkage analysis	PBE or DNA	5-10 ml	2-8 °C	60	Prescription/ medical impression/ reports	Y	Y	N
151	PKU Female	PKU (Phenylketonuria), PAH Gene, Germline Mutation, Diagnosis, Carrier Detection, Metabolic disorders	Mol- Gene Sequencing	Linkage analysis	PBE or DNA	5-10 ml	2-8 °C	30	Prescription/ medical impression/ reports	Y	Y	N
152	PKU Male	PKU (Phenylketonuria), PAH Gene, Germline Mutation, Diagnosis, Carrier Detection, Metabolic disorders	Mol- Gene Sequencing	Sanger Sequencing	PBE or DNA	5-10 ml	2-8 °C	30	Prescription/ medical impression/ reports	Y	Y	N
153	PKU Other	PKU (Phenylketonuria), PAH Gene, Germline Mutation, Diagnosis, Carrier Detection, Metabolic disorders	Mol- Gene Sequencing	Sanger Sequencing	PBE or DNA	5-10 ml	2-8 °C	30	Prescription/ medical impression/ reports	Y	Y	N



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154	PND- PGD	PGD-PGT-M-Monogenic disease-IVF-PND-Pre-PGT-MPreimplantation genetic diagnosis	Mol- PGD	Sanger Sequencing	CVS/Amnion	5-10 ml	2-8 °C	14	medical impression/ reports	Y	Y	N
155	PND-Alpha	PND- Alpha Thalassemia, Prenatal Diagnosis	Mol- Thalasemia	Sanger Sequencing- Multiplex PCR- Alpha MLPA	PBE or DNA or CV or AF or AFC	5-10 ml	2-8 °C	14	CBC- Hb Electrophoresis-Ferritin	Y	Y	Y
156	PND-CF	PND -CF (Cystic fibrosis), Sequencing, MLPA,Prenatal Diagnosis	Mol- Others	Sanger Sequencing/ Reverse Strip Assay	PBE or DNA	5-10 ml	2-8 °C	14	medical impression/ reports	Y	Y	Y
157	PND-Duchenne	PND -DMD , Sequencing, MLPA, Prenatal Diagnosis	Mol- Others	Sanger Sequencing/ML PA	PBE or DNA	5-10 ml	2-8 °C	14	medical impression/ reports	Y	Y	Y
158	PND-FraX	PND -FRAX, , PreMutation, Prenatal Diagnosis	Mol- Others	Fragment Analysis/Southe rn	PBE or DNA	5-10 ml	2-8 °C	14	medical impression/ reports	Y	Y	Y
159	PND-Gaucher	PND -Gaucher Germline Mutation, Prenatal Diagnosis	Mol- Others	Sanger Sequencing/ Reverse Strip Assay	PBE or DNA	5-10 ml	2-8 °C	14	medical impression/ reports	Y	Y	Y
160	PND-Mutation Confirmation	Germline Mutation, Sequencing, Prenatal Diagnosis	Mol- Gene Sequencing	Sanger Sequencing	PBE or DNA	5-10 ml	2-8 °C	14	Prescription/ medical impression/ reports	Y	Y	**Y



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161	PND-Others	PND, Prenatal Diagnosis	Mol- Others	Sanger Sequencing- MLPA	PBE or DNA	5-10 ml	2-8 °C	14	medical impression/ reports	Y	Y	Y
162	PND-PKU	PND-PKU, (Phenylketonuria), Germline Mutation, Prenatal Diagnosis, Metabolic disorders	Mol- Gene Sequencing	Sanger Sequencing	PBE or DNA	5-10 ml	2-8 °C	14	Prescription/ medical impression/ reports	Y	Y	**Y
163	PND-PRE	PND, NEW Variant	Mol- Gene Sequencing	Sanger Sequencing	PBE or DNA	5-10 ml	2-8 °C	60	Complete information of previous tests/clinical records/paraclinic tests	Y	Y	N
164	PND-Sex	PND- Sex Determination, Prenatal Diagnosis	Mol- Others	PCR	PBE or DNA	5-10 ml	2-8 °C	14	medical impression/ reports	Y	Y	Y
165	PND-Sickle	PND- Sickle, Prenatal Diagnosis	Mol- Thalasemia	Sanger Sequencing	PBE or DNA or CV or AF or AFC	5-10 ml	2-8 °C	14	CBC- Electrophoresis- Ferritin	Y	Y	Y
166	PND-SMA	PND, SMA, Prenatal Diagnosis	Mol- Others	MLPA	PBE or DNA	5-10 ml	2-8 °C	14	medical impression/ reports	Y	Y	Y
167	PND-Thal Beta	PND, Beta thalasemia, Prenatal Diagnosis	Mol- Thalasemia	Sanger Sequencing- Beta MLPA	PBE or DNA or CV or AF or AFC	5-10 ml	2-8 °C	14	CBC- Electrophoresis- Ferritin	Y	Y	Y
168	POLE Common Somatic Mutation	Somatic Mutatin, Endometrial Cancer POLE, Helisomatic mutations	Mol- Trans	Sanger Sequencing	Tis or DNA or FFPE	> 20% Tumoral Tissue	2-8 °C	14	medical compression/reports/path ology results/form TE- REX-FO-097/	Y	Y	N



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169	PT-Pro-Thrombin (Factor II)	Factor II, Prothrombin, PT	Mol- Others	Sanger Sequencing/ Reverse Strip Assay	PBE or DNA	5-10 ml	2-8 °C	14	medical impression/ reports	N	by the attending physician	N
170	QF-PCR Aneuploidies	Determination of Aneuploidy of Chr. 13, 18, 21, X and Y in fetus, abnormality, Screening, Aneuploidies	Mol- Trans	Fragment Analysis	PBE or DNA and AF or PBE or DNA and CVs or PBE or DNA and TC	3-5 ml Amnion or CVS and 5-10 ml PBE	2-8 °C	3	Prescription/ medical impression/ reports	Y	Y	N
171	RET Common Somatic mutation	Common Somatic Variant, Thyroid medullary Carcinoma, proto-oncogen, receptor tyrosin kinase	Mol- Trans	Sanger Sequencing	PBE or Tis or DNA	5-10 ml/> 20% Tumoral Tissue	2-8 °C	14	medical compression/reports/pathology results/prescription	N	by the attending physician	N
172	RT-PCR (1-19)	RT-PCR- ALL , Leukemia, Diagnosis	Mol- Trans	RT-PCR	PBE or BM	5-10 ml	2-8 °C	7	All disease diagnosis documents/ prescription / CBC	N	by the attending physician	N
173	RT-PCR (12-21)	RT-PCR- ALL , Leukemia, Diagnosis	Mol- Trans	RT-PCR/Real Time PCR	PBE or BM	5-10 ml	2-8 °C	7	All disease diagnosis documents/ prescription / CBC	N	by the attending physician	N
174	RT-PCR (15-17)	RT-PCR- AML, AML-M3, PML-RARA , Leukemia, Diagnosis	Mol- Trans	RT-PCR/Real Time PCR	PBE or BM	5-10 ml	2-8 °C	7	All disease diagnosis documents/ prescription / CBC	N	by the attending physician	N
175	RT-PCR (15-17) Quantitative PMLRARA	RT-PCR- AML, AML-M3, PML-RARA , Leukemia, Diagnosis	Mol- Trans	RT-PCR/Real Time PCR	PBE or BM	5-10 ml	2-8 °C	14	All disease diagnosis documents/ prescription / CBC	N	by the attending physician	N



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176	RT-PCR (4-11)	RT-PCR- ALL , Leukemia, Diagnosis	Mol- Trans	RT-PCR	PBE or BM	5-10 ml	2-8 °C	7	All disease diagnosis documents/ prescription / CBC	N	by the attending physician	N
177	RT-PCR (8-21)	RT-PCR- AML , Leukemia,Diagnosis	Mol- Trans	RT-PCR/Real Time PCR	PBE or BM	5-10 ml	2-8 °C	7	All disease diagnosis documents/ prescription / CBC	N	by the attending physician	N
178	RT-PCR (9-22) (P190KD-e1a2)	RT-PCR- CML, P190 BCR-ABL , Leukemia, Diagnosis	Mol- Trans	RT-PCR/Real Time PCR	PBE- BM	5-10 ml	2-8 °C	7	All disease diagnosis documents/ prescription / CBC	N	by the attending physician	N
179	RT-PCR (9-22) (P190KD- P210KD- P230KD)	RT-PCR- CML, P190 BCR-ABL , Leukemia, Diagnosis	Mol- Trans	RT-PCR/Real Time PCR	PBE- BM	5-10 ml	2-8 °C	7	All disease diagnosis documents/ prescription / CBC	N	by the attending physician	N
180	RT-PCR (9-22) (P210KD-b2a2- b3a2)	RT-PCR- CML, P210 BCR-ABL , Leukemia, Diagnosis	Mol- Trans	RT-PCR/Real Time PCR	PBE- BM	5-10 ml	2-8 °C	7	All disease diagnosis documents/ prescription / CBC	N	by the attending physician	N
181	RT-PCR (9-22) (P230KD-e19a2)	RT-PCR- CML, P210 BCR-ABL , Leukemia, Diagnosis	Mol- Trans	RT-PCR/Real Time PCR	PBE- BM	5-10 ml	2-8 °C	7	All disease diagnosis documents/ prescription / CBC	N	by the attending physician	N
182	RT-PCR (9-22) Quantitative (P190KD-e1a2)	RT-PCR- CML, P190 BCR-ABL , Leukemia, Diagnosis	Mol- Trans	RT-PCR/Real Time PCR	PBE- BM	5-10 ml	2-8 °C	14	All disease diagnosis documents/ prescription / CBC	N	by the attending physician	N
183	RT-PCR (9-22) Quantitative (P210KD-b2a2- b3a2)	RT-PCR- CML, P210 BCR-ABL , Leukemia, Diagnosis	Mol- Trans	RT-PCR/Real Time PCR	PBE- BM	5-10 ml	2-8 °C	14	All disease diagnosis documents/ prescription / CBC	N	by the attending physician	N



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184	RT-PCR inv (16)	RT-PCR- AML , Leukemia, Diagnosis	Mol- Trans	RT-PCR	PBE- BM	5-10 ml	2-8 °C	7	All disease diagnosis documents/ prescription / CBC	N	by the attending physician	N
185	Sequencing CAPN3	Sequencing, limb-girdle muscular dystrophy-1	Mol- Gene Sequencing	Sanger Sequencing	PBE or DNA	5-10 ml	2-8 °C	30	Prescription/ medical impression/ reports	Y	Y	N
186	Sequencing CF	CFTR-Cyctic Fibrosis- Germline Mutation, Sequencing, Diagnosis, Carrier Detection	Mol- Gene Sequencing	Sanger Sequencing	PBE or DNA	5-10 ml	2-8 °C	60	Prescription/ medical impression/ reports	Y	Y	N
187	Sequencing DMD/BMD	DMD, Duchenne muscular dystrophy - Germline Mutation, Sequencing, Diagnosis, Carrier Detection	Mol- Gene Sequencing	Sanger Sequencing	PBE or DNA	5-10 ml	2-8 °C	60	Prescription/ medical impression/ reports	Y	Y	N
188	Sequencing FMF	MEFV, FMF- Germline Mutation, Sequencing, Diagnosis, Carrier Detection	Mol- Gene Sequencing	Sanger Sequencing	PBE or DNA	5-10 ml	2-8 °C	30	Prescription/ medical impression/ reports	Y	Y	N
189	Sequencing Hematochromato sis	Germline Mutation, Sequencing, Diagnosis, Carrier Detection	Mol- Gene Sequencing	Sanger Sequencing	PBE or DNA	5-10 ml	2-8 °C	30	Prescription/ medical impression/ reports	Y	Y	N
190	Sequencing Reaction	Sequencing RUN	Mol- Sequencing	Sanger Sequencing	PCR Product	25UL	2-8 °C	3	لیبل نمونه روی میکروتیوپ، ارسال پرایمر و اطلاعات ژن و پرایمر در اکسل ارسالی	N	N	N



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191	Sequencing Run	Sequencing Run	Mol- Sequencing	Sanger Sequencing	PCR Product	25UL	2-8 °C	3	لیبل نمونه روی میکرونیوپ، ارسال پرایمر و اطلاعات ژن و پرایمر در اکسل ارسالی	N	N	N
192	Sequencing TP53	Li-fraumeni Syndrome, P53, TP53, Germline Mutation, Sequecing, Diagnosis, Tumer protein P53, TRP53	Mol- Gene Sequencing	Sanger Sequencing	PBE or DNA	5-10 ml	2-8 °C	30	Prescription/ medical impression/ reports	Y	Y	N
193	SEX- Determination	Sex-Determination	Mol- Others	PCR	PBE or DNA	5-10 ml	2-8 °C	5	medical impression/ reports	Y	Y	N
194	SF3B1 Common Somatic Mutation	Cancer,Sanger Sequencing, MDS، جهش های هلی سوماتیک، Breast Cancer, CLL, Endometrial Cancer	Mol- Trans	Sanger Sequencing	PBE or DNA	5-10 ml	2-8 °C	14	medical impression/ reports	N	Y	N
195	Sickle Female	Sikle cell Thalassemia	Mol- Thalasemia	Sanger Sequencing	PBE or DNA	5-10 ml	2-8 °C	14	CBC- Hb Electrophoresis-Ferritin	Y	N	N
196	Sickle Male	Sikle cell Thalassemia	Mol- Thalasemia	Sanger Sequencing	PBE or DNA	5-10 ml	2-8 °C	14	CBC- Hb Electrophoresis-Ferritin	Y	N	N
197	Sickle Other	Sikle cell Thalassemia	Mol- Thalasemia	Sanger Sequencing	PBE or DNA	5-10 ml	2-8 °C	14	CBC- Hb Electrophoresis-Ferritin	Y	N	N



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198	SLOS Gene Sequencing	Smith Lemli Opitz Syndrome, deficiency in the enzyme 7-dehydrocholesterol reductase, abnormality in cholesterol metabolism	Mol- Gene Sequencing	Sanger Sequencing	PBE or DNA	5-10 ml	2-8 °C	14	Prescription/ medical impression/ reports	Y	جواب غیر طبیعی	N
199	SMA Female	SMA-Spinal Muscular Atrophy, Diagnosis, Carrier Detection	Mol- Others	MLPA	PBE or DNA	5-10 ml	2-8 °C	14	medical impression/ reports	Y	جواب غیر طبیعی	N
200	SMA FraX- Carrier screening female	Reproductive Screening, Pre-Pregnancy, Germline Screening, Carrier Screening	Mol- Others	MLPA, TP-PCR	PBE or DNA	5-10 ml	2-8 °C	30	Prescription/ medical impression/ reports	Y	جواب غیر طبیعی	N
201	SMA Male	SMA-Spinal Muscular Atrophy, Diagnosis, Carrier Detection	Mol- Others	MLPA	PBE or DNA	5-10 ml	2-8 °C	14	medical impression/ reports	Y	جواب غیر طبیعی	N
202	SMA Other	SMA-Spinal Muscular Atrophy, Diagnosis, Carrier Detection	Mol- Others	MLPA	PBE or DNA	5-10 ml	2-8 °C	14	medical impression/ reports	Y	جواب غیر طبیعی	N
203	SRSF2 Common Somatic Mutation	SRSF2, Somatic Mutation, Myeloproliferative Neoplasms (MPN), Cancer, Pharmacogenetics	Mol- Trans	Sanger Sequencing	PBE or Tis or DNA or FFPE	5-10 ml/> 20% Tumoral Tissue	2-8 °C	14	medical compression/ reports/ pathology results/ doctor's prescription	Y	by the attending physician	N
204	SRY/Sex Determination	SRY, Sex Determination	Mol- Others	PCR	PBE or DNA	5-10 ml	2-8 °C	3	medical impression/ reports	Y	جواب غیر طبیعی	N





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205	TERT Common Somatic Mutation	TERT, Somatic Mutation, Nervous System Cancer, Cancer, Pharmacogenetics	Mol- Trans	Sanger Sequencing	Tis or DNA or FFPE	> 20% Tumoral Tissue	2-8 °C	14	medical compression/ reports/ pathology results/ doctor's prescription	Y	by the attending physician	*Y
206	Thal II-1-XMN	Thalassemia, XMN, Carrier Detection, Thalassemia Major	Mol- Thalasemia	ARMS PCR- XMNI Sanger Sequencing	PBE or DNA	5-10 ml	2-8 °C	14	CBC- Hb Electrophoresis	Y	N	N
207	TP53 Common Somatic Mutation	TP53, Somatic Mutation, Chronic Lymphocytic Leukemia (CLL), Myeloproliferative Neoplasms, Cancer, Pharmacogenetics	Mol- Trans	Sanger Sequencing	Tis or DNA or FFPE	> 20% Tumoral Tissue	2-8 °C	14	medical compression/ reports/ pathology results/ doctor's prescription	N	by the attending physician	N
208	U2AF1 Common Somatic Mutation	U2AF1, Common Somatic Mutation, Pharmacogenetics,, Myeloproliferative Neoplasms (MPN), Cancer	Mol- Trans	Sanger Sequencing	Tis or DNA or FFPE	> 20% Tumoral Tissue	2-8 °C	14	medical compression/ reports/ pathology results/ doctor's prescription	N	by the attending physician	*Y
209	Warfarin Resistance	Warfarin Resistance	Mol- Others	Reverse Strip Assay	PBE or DNA	5-10 ml	2-8 °C	14	medical impression/ reports	Y	by the attending physician	N
210	WGA PGD I		Mol- PGD	amplification	Blastomere/ blastocyst	1-4 Blastomeres	2-8 °C	2	medical impression/ reports	N	Y	Y
211	WGA PGD II		Mol- PGD	amplification	Blastomere/ blastocyst	>4 Blastomeres	2-8 °C	2	medical impression/ reports	N	Y	Y



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212	WT1 common somatic mutation	AML, ALL, Prostate cancer, Breast cancer	Mol- Trans	Sanger Sequencing	Tis or DNA or FFPE or PBE	5-10 ml	2-8 °C	14	medical compression/ reports/ pathology results/ doctor's prescription	N	Y	N
213	Y-Microdeletion	Y-Microdeletion; Male Infertility , Azoospermia	Mol- Others	PCR	PBE or DNA	5-10 ml	2-8 °C	14	medical impression/ reports	Y	Y	N