

مرکزپاتولوژی و ژنتیک کریمی نژاد- نجم آبادی

ردیف	Test	کلیدواژه keyword	ID	روش انجام method	نوع نمونه sample	حجم نمونه volume	شرایط نگهداری storage	زمان جوابدهی(روز) TAT (Day)	مدارک موردنیاز dacument	نیاز به مشاوره قبل از انجام آزمایش دارد؟ Pre test counseling	نیاز به مشاوره بعد از انجام آزمایش دارد؟ Post test counseling	قابلیت اورژانس دارد؟ Does it have an emergency 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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247	APOE Genotyping	Sanger Sequencing, Alzhemer disease, Cardiovascular genetics, SNP, rs429358, rs7412	Mol- Gene Sequencing	Sanger Sequencing	Blood(DNA)	5-10 ml	2-8 °C	30	medical impression/ reports	Y	Y	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 MutationNervous 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-Common mutation	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
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

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16	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
17	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
226	CF Common Mutation Female	CFTR,Common variants	Mol-Others	Reverse Strip Assay	DNA	5-10 ml	2-8 °C	14	medical impression/ reports	N	by the attending physician	N
18	CF Common Mutation Male	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	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
20	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
21	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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22	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
242	Comprehensive Leukemia Fusion Gene Panel	Leukemia, AML, ALL, Fusion, Translocation, PML-RARA t(15;17) , AML1-ETo t(8;21) , CBFB-MYH11 (inv16), t(1;19) , t(12;21) , t(4;11) , , t(11;19) , t(9;11) , t(6;11) , t(10;11) , t(9;22) ,	Mol-T.L	PCR	Blood/Bone Marrow	5-10 ml	2-8 °C	14	medical impression/ reports	Y	Y	N
23	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
24	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
25	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
26	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
27	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
28	DM1	DM1	Mol- Others	Fragment Analysis	PBE or DNA	5-10 ml	2-8 °C	30	medical impression/ reports	Y	Y	N

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29	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
30	DNA Collection-mouth swab	DNA-BucuAL	Mol-Extraction	DNA Extraction	Tis	2swab	2-8 °C	3	Prescription	Y	N	N
217	DNA-Extraction	DNA-Extraction	Mol-Extraction	DNA Extraction	PBE or DNA-Blood/Bone Marrow/Tis or FFPE/cvs/amnion	5-10 ml Blood or 20% Tumoral Tissue	2-8 °C	3	Prescription	Y	N	N
218	DNA-Extraction-Free	DNA-Extraction	Mol-Extraction	DNA Extraction	PBE or DNA-Blood/Bone Marrow/Tis or FFPE/cvs/amnion	5-10 ml Blood or 20% Tumoral Tissue	2-8 °C	3	Prescription	Y	N	N
31	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
33	DPYD (5-fu)	colorectal cancer , Solid tumor , IVS14+1G>A	Mol-Trans	Reverse Strip Assay	PBE or DNA	5-10 ml	2-8 °C	14	medical impression/ reports	N	by the attending physician	N
34	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

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35	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
36	ESR1 Common Somatic Mutation	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
39	FancA-Seq-MLPA	FancA. Fanconi anemi, Germline Mutation	Mol- Gene Sequencing	Sanger Sequencing/MLPA	PBE or DNA	5-10 ml	2-8 °C	60	medical impression/ reports	Y	Y	N
40	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	medical impression/ reports/Pathology Reports/prescription	Y	by the attending physician	N
41	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
42	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
43	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
44	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



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45	FraX-Mol	FraX, Premature Ovarian Failure (POF), X link Syndromic Mental Retardation, Diadnosis, Carier Detection	Mol- Others	PCR- Fragment Analysis/Southern	PBE or DNA	5-10 ml	2-8 °C	30	medical impression/ reports	Y	Y	N
46	Friedreich`s ataxia	FXN, GAA Expanation, Friedreich`s ataxia	Mol- Others	PCR- Fragment Analysis	PBE or DNA	5-10 ml	2-8 °C	30	medical impression/ reports	Y	Y	N
48	Gaucher	GBA gene;beta-glucocerebrosidase, Dagnosis, Carrier Detection	Mol- Gene Sequencing	Sanger Sequencing	PBE or DNA	5-10 ml	2-8 °C	14	medical impression/ reports	N	Y	N
219	Gene Seq >51 Exon	Gemline Mutation, 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
49	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
50	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
51	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
53	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

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54	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
55	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
56	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
15	Hereditary Diffuse Gastric Cancer (HDGC); CDH1	Gastric cancer, Sanger sequencing, Hereditary cancer	Mol-Gene seq	Sanger Sequencing	PBE	10 ml	2-8 °C	60	medical impression/ reports	Y	Y	N
256	Hereditary Diffuse Gastric Cancer (HDGC); CDH1 gene	CANCER/SOLIDT TUMOR	MOL-Genseq	SANGER SEQUNCING	blood	5-10 ml	2-8 °C	60	Prescription/ medical impression/ reports/clinical form/pathology report	Y	Y	N
57	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
58	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
59	IDH1 Common Somatic Mutation	IDH1, Somatic Mutation, Nervous system Cancer, Myeloprolifrative Neoplasms (MPN)	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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60	IDH2 Common Somatic Mutation	IDH2, Somatic Mutation, Nervous system Cancer, Myeloprolifrative Neoplasms (MPN)	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
61	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
62	Irinotecan Toxicity	UGT1A1*28 genotype, Pharmacogenomics, Pancreatic Neoplasms, Colorectal Neoplasms,	Mol- Trans	Sanger Sequencing	PBE or DNA	5-10 ml	2-8 °C	14	medical impression/ reports	N	جواب غیر طبیعی	N
63	JAK2 Exon 12	JAK2, Myeloprolifrative Neoplasms (MPN), Leukemia, MPN	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
220	JAK2 gene V617 Quantitive	JAK2, V617F, Leukemia, Myeloprolifrative Neoplasms (MPN),CML,AML	Mol-T.L	Real time PCR	PBE or DNA	5-10 ml	2-8 °C	7	CBC/ Prescription/ medical impression/ reports	N	by the attending physician	N
64	JAK2 gene v617F Mutation	JAK2, V617F, Leukemia, Myeloprolifrative Neoplasms (MPN),CML,AML	Mol- Trans	Sanger Sequencing/ Real time PCR	PBE or DNA	5-10 ml	2-8 °C	7	CBC/ Prescription/ medical impression/ reports	N	by the attending physician	N

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65	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
66	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
67	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
68	MLPA- CAPN3	MLPA- Large Deletion /Insertion, Diagnosis, Carrier Detection, CAPN3, CALpain3	Mol- Others	MLPA	PBE or DNA	5-10 ml	2-8 °C	30	Prescription/ medical impression/ reports	Y	Y	N
69	MLPA- Celiac DQ2 & DQ8	Celiac disease (HLA-DQ2/ HLA-DQ8)	Mol- Others	MLPA	PBE or DNA	5-10 ml	2-8 °C	30	Prescription/ medical impression/ reports	Y	N	N
70	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
71	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
72	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

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73	MLPA-BRCA I-II	MLPA, Breast and Ovarian Cancer, Diagnosis, Carrier Detection	Mol- Others	MLPA	PBE or DNA	5-10 ml	2-8 °C	30	medical impression/ reports	Y	Y	N
74	MLPA-CLL	deletion, Insertion, Chrionic lymphocytic leukemia	Mol- Trans	MLPA	PBE or DNA	5-10 ml	2-8 °C	14	Prescription/ medical impression/ reports	Y	Y	N
75	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
265	MLPA-CYP2D6	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
76	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
221	MLPA-Del/Dup II-...	MLPA- Large Deletion /Insertion, Diagnosis, Carrier Detection	Mol-Others	MLPA	PBE or DNA	5-10 ml	2-8 °C	14	medical impression/ reports	Y	Y	N
77	MLPA-MGMT Methylation	Hypermethylation, glioma	Mol- Trans	MLPA	Tis Or DNA or FFPE	> 20% Tumoral Tissue/5-10 ml	2-8 °C	30	medical impression/ reports/Pathology Reports/prescription	N	by the attending physician	Y

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78	MLPA-Microdeletions	MLPA- Deletion /Insertion, Diagnosis, Carrier Detection1p36 deletion syndrome/2p16.1-p15	Mol- Others	MLPA	PBE or DNA	5-10 ml	2-8 °C	30	medical impression/ reports	N	N	N
79	MLPA-MLH1-2,PMS2,MSH6 Hypermethylation	Hypermethylation, Somatic Mutation, Mis match repair gene, microsatellitc instability	Mol- Trans	MLPA	Tis Or DNA or FFPE	> 20% Tumoral Tissue/5-10 ml	2-8 °C	30	medical impression/ reports/Pathology Reports/prescription	Y	by the attending physician	N
240	MLPA-Pharmacode	MLP-Pharma	Mol-T.L	MLPA	Blood(DNA)	5-10 ml	2-8 °C	14	medical impression/ reports	N	N	N
81	MLPA-SGC	Limb-girdle muscular dystrophy; MLPA- Large Deletion Duplication, Diagnosis, Carrier Detection	Mol- Others	MLPA	PBE or DNA	5-10 ml	2-8 °C	30	medical impression/ reports	Y	Y	N
80	MLPA-Strip-Mutation CAH (CYP21A2)	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
222	Molecular Cancer Report	Molecular Cancer Report	Molecular	_	_	5-10 ml	_	_	_	_	_	_
82	MPL-Mutation	MPL, Somatic Mutation-Myeloprolifrative 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

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83	MSI	Micro Satellite Inastability- Colorectal Cancer , MMR, Miss Match Repare, 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
86	Mutation Confirmation	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
87	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
88	NGS- Carrier Screening- Medical Exome	Carrier Screening, OMIM Genes	Mol- NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	90	Prescription/ medical impression/ reports/pedigree	Y	Y	N
89	NGS- Multi Cancer With CNV Panel (Solid Tumer and Non solid Tumer Cancer)	Solid Tumer, Cancer, NGS, Germline, Somatic	Mol- NGS	NGS	PBE or DNA or Tis or FFPE	5-10 ml/> 20% Tumoral Tissue	2-8 °C	90	Prescription/ medical impression/ reports	Y	Y	N
91	NGS-Ataxia Exome	NGS, Ataxia, Gemline Mutation, Diagnosis, Carrier Detection	Mol- NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	90	Prescription/ medical impression/ reports	Y	Y	N
90	NGS-Autism Spectrum Disordes-Exome	Autism, ASD, intellectual disability	Mol- NGS	NGS	PBE or DNA or Tis or FFPE	5-10 ml/> 20% Tumoral Tissue	2-8 °C	90	Prescription/ medical impression/ reports	Y	Y	N



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92	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
94	NGS-BRCA I-II Plus del/dup (Somatic and Germline) 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
223	NGS-Cardiac Screening-Exome	NGS-Cardiac Screening-Exome	Mol-NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	90	Prescription/ medical impression/ reports	Y	Y	N
95	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	Prescription/ medical impression/ reports	Y	Y	N
96	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	60	Prescription/ medical impression/ reports	Y	Y	N



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97	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	Prescription/ medical impression/ reports	Y	Y	N
266	NGS-CARRIER-without DNA	NGS-CARRIER-without DNA	Mol- NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	60	Prescription/ medical impression/ reports	Y	Y	N
98	NGS-Ciliopathies Exome	NGS-Ciliopathies, Gemline Mutation, Diagnosis, Carrier Detection	Mol- NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	90	Prescription/ medical impression/ reports	Y	Y	N
224	NGS-Clinical Exome (Behzisti)	Gemline Mutation, Diagnosis, Carrier screening	Mol-NGS	NGS	PBE or Tis	5-10 ml	2-8 °C	90	Order/Previous genetic and paraclinic test results/Pedigree	Y	Y	N
267	NGS-Clinical Exome (Behzisti-zanjan)	Gemline Mutation, Diagnosis, Carrier screening	Mol-NGS	NGS	PBE or Tis	5-10 ml	2-8 °C	90	Order/Previous genetic and paraclinic test results/Pedigree	Y	Y	N
99	NGS-CMT & Sensory Neuropathies Exome	NGS, Sensory Neuropathies, Gemline Mutation, Diagnosis, Carrier Detection	Mol- NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	90	Prescription/ medical impression/ reports	Y	Y	N

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100	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	90	Prescription/ medical impression/ reports	Y	Y	N
101	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	Prescription/ medical impression/ reports	Y	Y	N
102	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	Prescription/ medical impression/ reports	Y	Y	N
103	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	Prescription/ medical impression/ reports	Y	Y	N
104	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	Prescription/ medical impression/ reports	Y	Y	N
105	NGS-Epilepcy Exome	NGS-Epilepcy, Gemline Mutation, Diagnosis, Carrier Detection	Mol- NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	90	Prescription/ medical impression/ reports	Y	Y	N
106	NGS-Exome (Medical exome for OMIM refrenced genes 200X)	NGS,Gemline Mutation, Diagnosis, Carrier Detection	Mol- NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	90	Prescription/ medical impression/ reports	Y	Y	N

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107	NGS-Exome (Medical exome for OMIM refrenced genes)	NGS-Exome (Medical exome for OMIM refrenced genes), Gemline Mutation, Diagnosis, Carrier Detection	Mol- NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	90	Prescription/ medical impression/ reports	Y	Y	N
108	NGS-Exome+ Mito 100X	Mitochondrial disrder, hetroplasmy, mt DNA Screening	Mol- NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	90	Prescription/ medical impression/ reports	Y	Y	N
246	NGS-Express-Carrier Screening-Medical Exome	NGS-Express-Carrier Screening-Medical Exome	Mol-NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	50	Prescription/ medical impression/ reports			
216	NGS-Express-Exome	NGS,Gemline Mutation, Diagnosis, Carrier screening, Detection	Mol-NGS	NGS	PBE or Tis	5-10 ml	2-8 °C	50	Order/Previous genetic and paraclinic test results/Pedigree	Y	Y	N
245	NGS-Express-Exome 200X	NGS-Express-Exome 200X	Mol-NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	50	Prescription/ medical impression/ reports	Y	Y	N
244	NGS-Express-Parents (Including carrier detection)-Exome	NGS-Express-Parents (Including carrier detection)-Exome	Mol-NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	50	Prescription/ medical impression/ reports	Y	Y	N
243	NGS-Express-Trio (Proband and 2 family preferred members)-Exome	NGS-Express-Trio (Proband and 2 family preferred members)-Exome	Mol-NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	50	Prescription/ medical impression/ reports	Y	Y	N

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227	NGS-Extra Analysis	Common variants, carrier screening	Mol-NGS	NGS	PBE or Tis	5-10 ml	2-8 °C	90	Order/Previous genetic and paraclinic test results/Pedigree	Y	Y	N
109	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	Prescription/ medical impression/ reports	Y	Y	N
110	NGS-Hemostatic/Thrombotic disorders Exome	NGS-Hemostatic/Thrombotic disorders, Gemline Mutation, Diagnosis, Carrier Detection	Mol- NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	90	Prescription/ medical impression/ reports	Y	Y	N
111	NGS-Hereditary Cancer	NGS,Hereditary Cancer, Gemline Mutation, Diagnosis, Carrier Detection	Mol-Gene seq	NGS	PBE or DNA	5-10 ml	2-8 °C	90	Prescription/ medical impression/ reports	Y	Y	N
112	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
114	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	Prescription/ medical impression/ reports	Y	Y	N
116	NGS-IBMFS (Aplastic anemia)	Aplastic anemia, Fanconi anemia, shmachman diamond syndrome, diamond blackfan syndrome, bone marrow failure	Mol-Gene seq	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

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115	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	Prescription/ medical impression/ reports	Y	Y	N
117	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	Prescription/ medical impression/ reports	Y	Y	N
118	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	Prescription/ medical impression/ reports	Y	Y	N
113	NGS-Mendelian Disorders-without DNA	NGS-Mendelian Disorders-without DNA	Mol- NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	60	Prescription/ medical impression/ reports	Y	Y	N
119	NGS-Metabolic disorders Exome	NGS-Metabolic disorders, Metabolic disorders	Mol- NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	90	Prescription/ medical impression/ reports	Y	Y	N
258	NGS-Metabolic disorders-Exome	NGS-Metabolic disorders, Metabolic disorders	Mol- NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	90	Prescription/ medical impression/ reports	Y	Y	N
120	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	Prescription/ medical impression/ reports	Y	Y	N
121	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	Prescription/ medical impression/ reports	Y	Y	N

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122	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	Prescription/ medical impression/ reports	Y	Y	N
123	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	Prescription/ medical impression/ reports	Y	Y	N
259	NGS-Neuromuscular-Exome	NGS_Neuromuscular	Mol- NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	90	Prescription/ medical impression/ reports	Y	Y	N
250	NGS-Onco Deep Panel, HRD, TMB, MSI, Fusions, Mutations	OncoDEEP/TMB/HRD/Fusion/SNV/CNV/MSI	Mol-T.L	NGS	Paraffin blocks	حداقل دو میکروتیوپ حاوی برش های بلوک پارافینی	2-8 °C	4-6 weeks	Prescription/ medical impression/ reports/clinical form/pathology report	Y	Y	N
251	NGS-Onco HRD	HRD, HRR genes(e.g BRCA1,2,…)	Mol-T.L	NGS	Paraffin blocks	حداقل یک میکروتیوپ حاوی برش های بلوک پارافینی	2-8 °C	4-6 weeks	Prescription/ medical impression/ reports	Y	Y	N
125	NGS-Onco Multi cancer RNA fusion panel	Leukemia,RNA,NGS,Cancer,RAF1,BRAF,NTRK1,23, EGFR,FGFR3,PBX1,NRG,ROS1,RET,ALK,TFE3,PRKACA,ERG,PPARG,MET	Mol-Trans	NGS	PBE or FFPE	10 ml	2-8 °C	30	Prescription/ medical impression/Pathology reports	Y	Y	N

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124	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-Trans	NGS	PBE or DNA	10 ml	2-8 °C	30	Prescription/ medical impression/ reports	Y	Y	N
249	NGS-Onco Myeloid Panel, PBE, BM, NGS, Cancer	Onco Myeloid Panel - NGS	Mol-T.L	NGS	PBE or DNA	10 ml	2-8 °C	30	Prescription/ medical impression/ reports	Y	Y	N
262	NGS-OncoSelect	Ct DNA /Blood	Mol-T.L	NGS	blood	دو لوله 10 سی سی در لوله های مخصوص Cell free	2-8 °C	4-6 weeks	Prescription/ medical impression/ reports/clinical form/pathology report	Y	Y	N
263	NGS-OncoXplore	Ct DNA	Mol-T.L	NGS	blood	دو لوله 10 سی سی در لوله های مخصوص Cell free	2-8 °C	4-6weeks	Prescription/ medical impression/ reports/clinical form/pathology report	Y	Y	
126	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	Prescription/ medical impression/ reports	Y	Y	N
127	NGS-Primary immundeficincies Exome	NGS-Primary immundeficincies, Gemline Mutation, Diagnosis, Carrier Detection	Mol- NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	90	Prescription/ medical impression/ reports	Y	Y	N

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228	NGS-Raw Data (Fastq/VCF)	Gemline Mutation, Diagnosis, Carrier screening	Mol-NGS	NGS data analysis	FASTQ file	.	.	60	Order/Previous genetic and paraclinic test results/Pedigree	Y	Y	N
128	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
229	NGS-Recurrent Pregnancy Loss-Exome	Pregnancy loss, Carrier screening	Mol-NGS	NGS	PBE	5-10 ml	2-8 °C	90	Order/Previous genetic and paraclinic test results/Pedigree	Y	Y	N
129	NGS-Research Exome 100X	NGS,Research, Gemline Mutation, Diagnosis, Carrier Detection	Mol- NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	90	Prescription/ medical impression/ reports	Y	Y	N
130	NGS-RP Exome	NGS,Gemline Mutation, Diagnosis, Carrier Detection	Mol- NGS	NGS	PBE or DNA	5-10 ml	2-8 °C	90	Prescription/ medical impression/ reports	Y	Y	N
131	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	Prescription/ medical impression/ reports	Y	Y	N
132	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
133	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	Prescription/ medical impression/ reports	Y	Y	N

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134	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	Prescription/ medical impression/ reports	Y	Y	N
135	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
136	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
137	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
138	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
139	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
140	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

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141	Paternity	Paternity	Mol- Trans	Fragment Analysis	PBE or DNA	5-10 ml	2-8 °C	14	Identity documents	Y	Y	N
142	Paternity-Ind	Paternity	Mol- Trans	Fragment Analysis	PBE or DNA	5-10 ml	2-8 °C	14	Identity documents	Y	Y	N
143	PCR-Ampilification	PCR	Mol- Trans	PCR	DNA in streck tubes	5-10 ml	2-8 °C	3	Prescription/ medical impression/ reports	N	Y	N
144	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
145	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
146	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
147	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
148	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

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149	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
150	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
230	PGD-Pre common genes	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
231	Pharma Code	drug response/cancer	Mol-T.L	NGS	Blood(DNA)	5-10cc	2-8 °C	60	Prescription/ medical impression/ reports	N	by the attending physician	N
152	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
153	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

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154	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
155	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
156	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
157	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
158	PND-Duchenne	PND -DMD , Sequencing, MLPA, Prenatal Diagnosis	Mol- Others	Sanger Sequencing/MLPA	PBE or DNA	5-10 ml	2-8 °C	14	medical impression/ reports	Y	Y	Y
159	PND-FraX	PND -FRAX, , PreMutation, Prenatal Diagnosis	Mol- Others	Fragment Analysis/Southern	PBE or DNA	5-10 ml	2-8 °C	14	medical impression/ reports	Y	Y	Y
160	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

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161	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
162	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
163	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
164	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
165	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
166	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
167	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
168	PND-Thal Beta	PND, Beta thalassemia,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
169	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/pathology results/form TE-REX-FO-097/	Y	Y	N

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171	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
172	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
232	RNA-cDNA	RNA-cDNA	Mol-T.L	cDNA synthesis	RNA	5-10 ml	2-8 °C	_	_	_	_	_
233	RNA-Extraction	RNA-Extraction	Mol-Extraction	RNA-Extraction	PBE or FFPE	5-10 ml Blood or 20% Tumoral Tissue	2-8 °C	3	medical impression/ reports	Y	N	N
234	RNA-Extraction-Free	RNA-Extraction-Free	Mol-Extraction	RNA-Extraction	PBE or FFPE	5-10 ml Blood or 20% Tumoral Tissue	2-8 °C	3	medical impression/ reports	Y	N	N
270	RNA-Extraction-Report	RNA-Extraction-Report	Mol-Extraction	RNA-Extraction-Report	PBE or FFPE	5-10 ml Blood or 20% Tumoral Tissue	2-8 °C	3	medical impression/ reports	Y	N	N

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173	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
174	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
175	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
176	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
177	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
178	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
179	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

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180	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
181	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
182	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
183	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
184	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
185	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
238	SCA Repeats Panel	SCA, ATXN,DRPLA, ATN1,CACNA1A	Mol-Others	TP- PCR	PBE or DNA	5-10 ml	2-8 °C	30	medical impression/ reports	N	N	N
241	SCA Repeats-Ind	SCA, ATXN,DRPLA, ATN1,CACNA1A	Mol-Others	TP- PCR	PBE or DNA	5-10 ml	2-8 °C	30	medical impression/ reports	N	N	N
151	Sequencing AGXT	Sequencing	Mol-GeneSeq	Sanger sequencing	DNA	5-10 ml	2-8 °C	30	Prescription/ medical impression/ reports	Y	Y	N

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186	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
187	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
188	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
189	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
190	Sequencing Hematochromatosis	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
191	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
192	Sequencing Run	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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193	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
194	SEX-Determination	Sex-Determination	Mol- Others	PCR	PBE or DNA	5-10 ml	2-8 °C	5	medical impression/ reports	Y	Y	N
195	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
196	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
197	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
198	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
199	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
200	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

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201	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
202	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
203	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
254	Somatic-DNA-Extraction-Report	Somatic /FFPE/DNA	Mol-T.L	Salting out	Paraffin blocks	FFPE with tumoral cells	2-8 °C	5	Prescription/ medical impression/ reports/clinical form/pathology report	N	N	N
255	Somatic-DNA-Extraction-Report-Free	Somatic /FFPE/DNA	Mol-T.L	Salting out	Paraffin blocks	FFPE with tumoral cells	2-8 °C	5	Prescription/ medical impression/ reports/clinical form/pathology report	Y	Y	N
206	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
207	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

ردیف	Test	کلیدواژه keyword	ID	روش انجام method	نوع نمونه sample	حجم نمونه volume	شرایط نگهداری storage	زمان جوابدهی(روز) TAT (Day)	مدارک موردنیاز dacument	نیاز به مشاوره قبل از انجام آزمایش دارد؟ Pre test counseling	نیاز به مشاوره بعد از انجام آزمایش دارد؟ Post test counseling	قابلیت اورژانس دارد؟ Does it have an emergency function
208	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
209	TPMT	thiopurine therapy, thiopurine methyltransferase	Mol- Trans	Sanger Sequencing	PBE	5-10 ml	2-8 °C	14	medical impression/ reports	N	by the attending physician	N
235	Variant Reclassification (up to 3 variants)	Sanger Sequencing,NGS,VUS	Mol-GeneSeq	ACMG Analysis	–	–	–	3	medical impression/ reports	Y	Y	N
236	Variant Reclassification-Ind (Single variant)	VUS,NGS,Exome,Variant, Sanger Sequencing	Mol-GeneSeq	ACMG Analysis	–	–	–	3	medical impression/ reports	Y	Y	N
211	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
212	WGA I	whole genome amplification	Mol- PGD	amplification	Blastomere/ blastocyst	1-4 Blastomeres	2-8 °C	2	medical impression/ reports	N	Y	Y
213	WGA II	whole genome amplification	Mol- PGD	amplification	Blastomere/ blastocyst	>4 Blastomeres	2-8 °C	2	medical impression/ reports	N	Y	Y
214	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

مرکزپاتولوژی و ژنتیک کریمی نژاد- نجم آبادی

ردیف	Test	کلیدواژه keyword	ID	روش انجام method	نوع نمونه sample	حجم نمونه volume	شرایط نگهداری storage	زمان جوابدهی(روز) TAT (Day)	مدارک موردنیاز dacument	نیاز به مشاوره قبل از انجام آزمایش دارد؟ Pre test counseling	نیاز به مشاوره بعد از انجام آزمایش دارد؟ Post test counseling	قابلیت اورژانس دارد؟ Does it have an emergency function
237	WT1-MRD	AML, ALL,MRD	Mol-T.L	Real Time PCR	Blood/Bone Marrow/RNA	5-10 ml	2-8 °C	14	Prescription/ medical impression/ reports	N	by the attending physician	N
260	Y-Microdeletion (MLPA), High Resolution	Y-Microdeletion; Male Infertility , Azoospermia	Mol- Others	PCR	PBE or DNA	5-10 ml	2-8 °C	14	medical impression/ reports	Y	Y	N