

Test/Gene	Sample	Amount of Sample	T.A.T (day)	Method
CYTOGENETICS				
Chromosome analysis - Blood	WB (LH)	2-3ml	15	Karyotyping
Chromosome analysis - Amniotic fluid	AF	15-20ml	18-21	Karyotyping
Chromosome analysis - Amniotic fluid (tween)	AF	15-20ml	18-21	Karyotyping
Chromosome analysis - Chorionic villi	CVS	30-50µg	18-21	Karyotyping
Chromosome analysis - cord	Cord Blood (LH)	2-3ml	7	Karyotyping
Chromosome analysis - abortion	Abortion tissue	30-50µg	21	Karyotyping
Chromosome analysis - Bone marrow	BM (EDTA)	30-50µg	21	Karyotyping
Chromosome analysis + rapid FISH - AF	AF	15-20ml	2-3 + 21	Kary. + FISH
Chromosome analysis + rapid FISH - CVS	CVS	30-50µg	2-3 + 21	Kary. + FISH
MOLECULAR CYTOGENETICS (FISH)				
FISH - rapid FISH (13, 18, 21, X and Y)	AF/CVS	2-3ml	3	FISH analysis
FISH - Sperm FISH (13, 18, 21, X and Y)	Concentrated sperm	2-3ml	15	FISH analysis
FISH - DNA fragmentation (Tunel)	Concentrated sperm	2-3ml	15	FISH analysis
Microdeletion FISH - Alagille	WB (LH)/AF/CVS	2-3ml	7	FISH analysis
Microdeletion FISH - Cri-du chat	WB (LH)/AF/CVS	2-3ml	7	FISH analysis
Microdeletion FISH - Di-George/VCFS	WB (LH)/AF/CVS	2-3ml	7	FISH analysis
Microdeletion FISH - IGF1R	WB (LH)/AF/CVS	2-3ml	7	FISH analysis
Microdeletion FISH - Kallmann	WB (LH)/AF/CVS	2-3ml	7	FISH analysis
Microdeletion FISH - NF1	WB (LH)/AF/CVS	2-3ml	7	FISH analysis
Microdeletion FISH - Prader-Willi/Angelman	WB (LH)/AF/CVS	2-3ml	7	FISH analysis
Microdeletion FISH - Smith-Magenis	WB (LH)/AF/CVS	2-3ml	7	FISH analysis
Microdeletion FISH - Sotos	WB (LH)/AF/CVS	2-3ml	7	FISH analysis
Microdeletion FISH - Williams	WB (LH)/AF/CVS	2-3ml	7	FISH analysis
Microdeletion FISH - Wolf-Hirschhorn	WB (LH)/AF/CVS	2-3ml	7	FISH analysis
Microdeletion FISH - XIST	WB (LH)/AF/CVS	2-3ml	7	FISH analysis
MOLECULAR GENETICS				
ACE mutation analysis (I/D)	BS/WB (EDTA)	2-3ml	10	sequencing

Achondroplasia - FGFR3 (exon 10)	BS/WB (EDTA)/AF/CVS	2-3ml	10	sequencing
Achondroplasia - FGFR3 (exon 9,10,13,15)	BS/WB (EDTA)/AF/CVS	2-3ml	15	sequencing
Adenozin Deaminaz Def. - ADA (whole gene)	WB (EDTA)	2-3ml	30	sequencing
Alpha Thalassemia - HBA1&HBA2 (del.&dupl.)	BS/WB (EDTA)/AF/CVS	2-3ml	21	sequencing
Alpha-1-Antitrypsin Deficiency (AAT)	BS/WB (EDTA)/AF/CVS	2-3ml	10	Real time
ApoE (e2, e3 and e4 alleles)	BS/WB (EDTA)	2-3ml	10	Real time
Beta Thalassemia - HBB (whole gene)	BS/WB (EDTA)/AF/CVS	2-3ml	15	sequencing
Beta-Fibrinogen mutation analysis (-455G>A)	BS/WB (EDTA)	2-3ml	10	sequencing
Blackfan-Diamond Anemia - RPS19 (whole gene)	WB (EDTA)/AF/CVS	2-3ml	30	sequencing
CADAFIL - NOTCH3 (exon 3,4)	BS/WB (EDTA)	2-3ml	15	sequencing
CADAFIL - NOTCH3 (exon 3,4,11,18,19)	BS/WB (EDTA)	2-3ml	30	sequencing
Canavan - ASPA DNA analysis (whole gene)	WB (EDTA)/AF/CVS	2-3ml	30	sequencing
Cardiofaciocutaneous - BRAF (exon 6, 11-16)	BS/WB (EDTA)/AF/CVS	2-3ml	30	sequencing
Cardiofaciocutaneous - MEK1 (exon 2,3)	BS/WB (EDTA)/AF/CVS	2-3ml	15	sequencing
Cardiofaciocutaneous - MEK2 (exon 1, 2 and 3)	BS/WB (EDTA)/AF/CVS	2-3ml	15	sequencing
Cardiovascular risk panel - 12 mutation	BS/WB (EDTA)	2-3ml	15	sequencing
Celiak Disease – HLA (DQ2 and DQ8)	BS/WB (EDTA)/AF/CVS	2-3ml	10	Jel
Charcot-Marie-Tooth - MPZ (whole gene)	BS/WB (EDTA)/AF/CVS	2-3ml	30	sequencing
Charcot-Marie-Tooth (CMT1A) - PMP22 (whole gene)	BS/WB (EDTA)/AF/CVS	2-3ml	30	sequencing
Charcot-Marie-Tooth (CMT1A) - PMP22 dupl.	BS/WB (EDTA)/AF/CVS	2-3ml	21	fragment
Cold-induced sweating Syn. 1 - CRLF1 (whole gene)	BS/WB (EDTA)/AF/CVS	2-3ml	30	sequencing
Congenital Deafness - Conneksin 26 (whole gene)	BS/WB (EDTA)/AF/CVS	2-3ml	15	sequencing
Congenital Deafness - GJB1 (whole gene)	BS/WB (EDTA)/AF/CVS	2-3ml	15	sequencing
Cystic fibrosis - CFTR (19 mutation)	BS/WB (EDTA)/AF/CVS	2-3ml	10	sequencing
Cystic fibrosis - CFTR (36 mutation)	BS/WB (EDTA)/AF/CVS	2-3ml	10	sequencing
Cystic fibrosis - CFTR (550 mutation)	WB (EDTA)/AF/CVS	2-3ml	15	sequencing
Cystic fibrosis - CFTR (del&dupl.)	BS/WB (EDTA)/AF/CVS	2-3ml	21	fragment
Cystic fibrosis - CFTR (deltaF508)	BS/WB (EDTA)/AF/CVS	2-3ml	7	sequencing
Cystic fibrosis - CFTR (whole gene)	WB (EDTA)/AF/CVS	2-3ml	21	sequencing
Distonia (DYT1) – TOR1A1 (GAG deletion)	BS/WB (EDTA)/AF/CVS	2-3ml	21	sequencing
DMD/BM (EDTA)D - Dystrophin (79 exon del.&dupl.)	BS/WB (EDTA)/AF/CVS	2-3ml	21	fragment
Fabry - GLA (whole gene)	WB (EDTA)/AF/CVS	2-3ml	30	sequencing
Faktör II (prothrombin) mutation analysis	BS/WB (EDTA)	2-3ml	7	sequencing
Faktör V Leiden mutation analysis	BS/WB (EDTA)	2-3ml	7	sequencing
Faktör XII (HAE) mutation analysis	BS/WB (EDTA)	2-3ml	10	sequencing
Faktör XIII mutation analysis (V34L)	BS/WB (EDTA)	2-3ml	10	sequencing
FMF - MEFV (12 mutation))	BS/WB (EDTA)	2-3ml	10	sequencing
FMF - MEFV (122 mutation)	BS/WB (EDTA)	2-3ml	12	sequencing
FMF - MEFV (50 mutation)	BS/WB (EDTA)	2-3ml	15	sequencing
G6PD Deficiency - G6PD (whole gene)	WB (EDTA)/AF/CVS	2-3ml	30	sequencing
Galactocinase - GALK1 (whole gene)	WB (EDTA)/AF/CVS	2-3ml	30	sequencing
Galactosemia DNA analysis - GALT (whole gene)	WB (EDTA)/AF/CVS	2-3ml	30	sequencing
Gaucher Disease - GBA (whole gene)	WB (EDTA)/AF/CVS	2-3ml	30	sequencing
GSD Type 1A (Von Gierke) - G6PC (whole gene)	WB (EDTA)/AF/CVS	2-3ml	30	sequencing
GSD Type 1B/1C - SLC37A4 (whole gene)	WB (EDTA)/AF/CVS	2-3ml	30	sequencing
Hemochromatosis - H63D and C282Y	BS/WB (EDTA)	2-3ml	10	sequencing
HLAB27	BS/WB (EDTA)	2-3ml	10	Jel
HPA1 DNA analysis (a/b)	BS/WB (EDTA)	2-3ml	10	sequencing
Hunter Syndrome - IDUA (whole gene)	WB (EDTA)	2-3ml	30	sequencing
Hurler Syndrome - IDS (whole gene)	WB (EDTA)	2-3ml	30	sequencing

Hypochondroplasia - FGFR3 (exon 10)	BS/WB (EDTA)/AF/CVS	2-3ml	10	sequencing
Hypochondroplasia - FGFR3 (exon 9,10,13,15)	BS/WB (EDTA)/AF/CVS	2-3ml	15	sequencing
Hypoparat.-deafness-renal disp. - GATA3 (whole gene)	WB (EDTA)/AF/CVS	2-3ml	30	sequencing
LHON - MTND genes (A52T, R340H,M64V)	BS/WB (EDTA)/AF/CVS	2-3ml	30	sequencing
Maternal Contamination Testing	WB (EDTA)/AF/CVS	2-3ml	15	fragment
Metachromatic Lokodistrofi - ARSA (whole gene)	WB (EDTA)	2-3ml	30	sequencing
MTHFR (A1298C) mutation analysis	BS/WB (EDTA)	2-3ml	7	Real time
MTHFR (C677T) mutation analysis	BS/WB (EDTA)	2-3ml	7	Real time
Myotonic Dystrophy - DMPK (CTG tekrarı)	BS/WB (EDTA)	2-3ml	15	fragment
Neutropenia - ELANE (ELA2) (whole gene)	WB (EDTA)/AF/CVS	2-3ml	30	sequencing
Niemann-Pick - SMPD1 (whole gene)	WB (EDTA)/AF/CVS	2-3ml	30	sequencing
Osteopetrosis - CLCN7 (whole gene)	WB (EDTA)/AF/CVS	2-3ml	45	sequencing
Osteoporosis panel - 6 polymorphism	BS/WB (EDTA)	2-3ml	10	Chip array
PAI-1 DNA analysis (4G/5G)	BS/WB (EDTA)	2-3ml	10	sequencing
Rett Syndrome - MECP2 (del.&dupl.)	BS/WB (EDTA)/AF/CVS	2-3ml	30	sequencing
Rett Syndrome - MECP2 (whole gene)	BS/WB (EDTA)/AF/CVS	2-3ml	30	sequencing
Shwachman -Diamond Syndrome - SBDS (whole gene)	WB (EDTA)/AF/CVS	2-3ml	30	sequencing
Sialidosis (Nörominidaz) - Neu1 (whole gene)	WB (EDTA)/AF/CVS	2-3ml	30	sequencing
Sickle Cell Anemia	BS/WB (EDTA)/AF/CVS	2-3ml	10	sequencing
SMA carrier testing - SMN1	BS/WB (EDTA)/AF/CVS	2-3ml	21	fragment
Smith-Lemli-Opitz - DHCR7 (whole gene)	BS/WB (EDTA)/AF/CVS	2-3ml	45	sequencing
Spinal Muscular Atrophy (SMA) - SMN1	BS/WB (EDTA)/AF/CVS	2-3ml	21	fragment
Thanatophoric Dysplasia - FGFR3 (exon 10, 15)	BS/WB (EDTA)/AF/CVS	2-3ml	10	sequencing
Thrombophilia panel (factor II/factor V/MTHFR)	BS/WB (EDTA)/AF/CVS	2-3ml	7	sequencing
Y Chromosome Microdeletion - 20 STR	BS/WB (EDTA)/AF/CVS	2-3ml	10	Jel
PHARMACOGENETICS				
Drug sensitivity panel - extensive (28 polymorphism) CYP2D6, CYP2C9, CYP2C19, NAT2, CYP1A2	BS/WB (EDTA)	2-3ml	30	sequencing
Drug sensitivity panel - Standart (23 polymorphism) CYP2D6, CYP2C9 and CYP2C19	BS/WB (EDTA)	2-3ml	30	sequencing
Drug sensitivity panel (Neuropsychiatry) (24polymorphism) CYP2D6/CYP2C9/CYP2C19/CYP1A2/CYP3A4	BS/WB (EDTA)	2-3ml	30	sequencing
Coumadin Dose Panel - 3 polymorphism CYP2C9 and VKORC1	BS/WB (EDTA)	2-3ml	15	Real time
Plavix Dose Panel - 2 polymorphism CYP2C19 - 2 polymorphism	BS/WB (EDTA)	2-3ml	15	Real time
VKORC1 - 1639C>A	BS/WB (EDTA)	2-3ml	15	Real time
CYP2C9 - 2 polymorphism	BS/WB (EDTA)	2-3ml	15	Real time
CYP2C19 - 2 polymorphism	BS/WB (EDTA)	2-3ml	15	sequencing
CYP2C19 - 4 polymorphism	BS/WB (EDTA)	2-3ml	15	sequencing
CYP2D6 - 15 polymorphism	BS/WB (EDTA)	2-3ml	21	sequencing
CYP1A2 - 2 polymorphism	BS/WB (EDTA)	2-3ml	15	sequencing
CYP3A4 - 1 polymorphism	BS/WB (EDTA)	2-3ml	15	sequencing
5HTT - SSRI response	BS/WB (EDTA)	2-3ml	30	sequencing
UGT1A1 - *28 (İrinotekan)	BS/WB (EDTA)	2-3ml	15	sequencing
DPD - *2A (Fluorouracil - 5FU)	BS/WB (EDTA)	2-3ml	15	sequencing

TMPT - 4 polymorphism (Thiopurine)	BS/WB (EDTA)	2-3ml	15	sequencing
GSTM1 - 3 polymorphism	BS/WB (EDTA)	2-3ml	15	sequencing
HEMATO&ONCOLOGY				
ABL1 DNA analysis (exon 4-10)	WB (EDTA)/BM (EDTA) (EDTA)	2-3ml	15	sequencing
ABL1 DNA analysis (T315I)	WB (EDTA)/BM (EDTA)	2-3ml	10	sequencing
BRAF DNA analysis (exon 11 and 15)	TT		10	sequencing
BRAF DNA analysis (exon 15)	TT		10	sequencing
BRCA1&BRCA2 (whole gene)	WB (EDTA)	4-5ml	90	sequencing
CHEK2 DNA analysis (exon 10)	BS/WB (EDTA)	2-3ml	15	sequencing
Chimerism (FISH)	WB (LH)/BM (EDTA)	2-3ml	7	FISH analysis
Chimerism (PCR)	BS + WB (EDTA)	2-3ml	10	fragment
Chromosome analysis from bone marrow	BM (LH)	2-3ml	14	Karyotip
c-KIT DNA analysis - 2 exon	TT	2-3ml	10	sequencing
c-KIT DNA analysis - one exon	TT		10	sequencing
c-KIT DNA analysis (exon 9,11,13,17,18)	TT		15	sequencing
EGF DNA analysis (exon 1)	BS/WB (EDTA)	2-3ml	10	sequencing
EGFR DNA analysis (exon 18-21)	TT		15	sequencing
FISH - 1p36 & 19q13.3	TT		10	FISH analysis
FISH - 20q	WB (LH)/BM (LH)	2-3ml	7	FISH analysis
FISH - 20q13 (AURKA)	TT		10	FISH analysis
FISH - 5q deletion (EGR1 - 5q31)	WB (LH)/BM (LH)	2-3ml	7	FISH analysis
FISH - 7q deletion (D7S486 - 7q31)	WB (LH)/BM (LH)	2-3ml	7	FISH analysis
FISH - ALK (2p23)	TT	2-3ml	7	FISH analysis
FISH - AML1/ETO t(8;21)	WB (LH)/BM (LH)	2-3ml	7	FISH analysis
FISH - API2/MALT t(11;18)	WB (LH)/BM (LH)	2-3ml	7	FISH analysis
FISH - ATM (11q22.3)	WB (LH)/BM (LH)	2-3ml	7	FISH analysis
FISH - BCR gen lokusu (22q)	WB (LH)/BM (LH)	2-3ml	7	FISH analysis
FISH - BCR/ABL - t(9;22)	WB (LH)/BM (LH)	2-3ml	7	FISH analysis
FISH - CFBF - t(16;16), inv(16)	WB (LH)/BM (LH)	2-3ml	7	FISH analysis
FISH - cMET (7q31)	TT		10	FISH analysis
FISH - C-myc (8q24)	WB (LH)/BM (LH)	2-3ml	7	FISH analysis
FISH - D13S319 (13q14.3)	WB (LH)/BM (LH)	2-3ml	7	FISH analysis
FISH - E2A (TCF3)(19p13.3)	WB (LH)/BM (LH)	2-3ml	7	FISH analysis
FISH - EGFR (7p11.1-q11.1)	TT		10	FISH

				analysis
FISH - EWS1 (22q12)	TT		10	FISH analysis
FISH - FIP1L1/PDGFR A	WB (LH)/BM (LH)	2-3ml	7	FISH analysis
FISH - Her2/Neu (17q11-12)	TT		10	FISH analysis
FISH - IGF1R (15q26)	TT		10	FISH analysis
FISH - IGH/BCL2 t(14;18)	WB (LH)/BM (LH)	2-3ml	7	FISH analysis
FISH - IGH/CCND1 t(11;14)	WB (LH)/BM (LH)	2-3ml	7	FISH analysis
FISH - IGH/FGFR3 t(4;14)	WB (LH)/BM (LH)	2-3ml	7	FISH analysis
FISH - IGH/MAF t(14;16)	WB (LH)/BM (LH)	2-3ml	8	FISH analysis
FISH - IGH/MYC t(8;14)	WB (LH)/BM (LH)	2-3ml	7	FISH analysis
FISH - Kromozom 12	WB (LH)/BM (LH)	2-3ml	7	FISH analysis
FISH - Kromozom 8	WB (LH)/BM (LH)	2-3ml	7	FISH analysis
FISH - LAMP1 (13q34)	WB (LH)/BM (LH)	2-3ml	7	FISH analysis
FISH - MALT1 (18q21)	WB (LH)/BM (LH)	2-3ml	7	FISH analysis
FISH - MLL (11q23)	WB (LH)/BM (LH)	2-3ml	7	FISH analysis
FISH - N-myc (2p24)	TT/WB (LH)/BM (LH)	2-3ml	10	FISH analysis
FISH - p53 (17p13.1)	TT/WB (LH)/BM (LH)	2-3ml	10	FISH analysis
FISH - PML/RARA - t(15;17)	WB (LH)/BM (LH)	2-3ml	7	FISH analysis
FISH - RARA (17q21)	WB (LH)/BM (LH)	2-3ml	7	FISH analysis
FISH - RB1 (13q14)	WB (LH)/BM (LH)	2-3ml	10	FISH analysis
FISH - TEL/AML1 t(12;21)	WB (LH)/BM (LH)	2-3ml	7	FISH analysis
FISH - TOP2A (17q21-q22)	TT/WB (LH)/BM (LH)		10	FISH analysis
FLT3 - ITD and D835	WB (EDTA)/BM (EDTA)	2-3ml	15	sequencing
HRAS DNA analysis (exon 1 and 2)	TT	2-3ml	10	sequencing
IDH1 DNA analysis (exon 4)	WB (EDTA)	2-3ml	15	sequencing
IDH2 DNA analysis (exon 4)	WB (EDTA)	2-3ml	15	sequencing
JAK2 - V617F mutation	WB (EDTA)/BM (EDTA)	2-3ml	10	Real time
Kras DNA analysis (exon 1)	TT		10	sequencing
Kras DNA analysis (exon 1, 2 and 3)	TT		15	sequencing
Kras DNA analysis (whole gene)	TT	2-3ml	15	sequencing
MAPK1 DNA analysis (ERK2-p42) (exon 7)	WB (EDTA)	2-3ml	10	sequencing
Microsatellit Instability (MSI)	BS or WB (EDTA) + TT	2-3ml	15	fragment

MPL DNA analysis (exon 10)	WB (EDTA)/BM (EDTA)	2-3ml	15	sequencing
NPM1 DNA analysis (exon 12)	WB (EDTA)/BM (EDTA)	2-3ml	15	sequencing
NRAS DNA analysis (exon 1 and 2)	TT	2-3ml	15	sequencing
P16 (CDKN2A) - whole gene	TT	2-3ml	15	sequencing
P53 DNA analysis (exon 4-9)	BS /TT/WB (EDTA)	2-3ml	21	sequencing
PDGFRA (exon 12, 18 and 19)	TT	2-3ml	15	sequencing
PDGFRB (exon 18 and 19)	TT		15	sequencing
PIK3CA DNA analysis (exon 9 and 20)	TT/WB (EDTA)	2-3ml	15	sequencing
PTEN DNA analysis (whole gene)	BS /TT/WB (EDTA)	2-3ml	30	sequencing
Real time - BCR/ABL t(9;22)	TT/WB (EDTA)	2-3ml	10	Real time
Real time - PML/RARA t(15;17)	WB (EDTA)	2-3ml	10	Real time
Ret (exon 10,11,13-16)	BS /WB (EDTA)	2-3ml	21	sequencing
MOLECULAR MICROBIOLOGY				
HPV screening + typing (35 different types)	ThinPrep/ Paraffinised tissue		7	Chip array

The following are abbreviation explaining the tissue needed or the tube for the test:

BS: buccal swap	AF: amniotic fluid	LH: lithium heparin
BM (EDTA): BM (EDTA)	CVS: chorionic villi	
WB: whole blood	TT: tumour tissue (fresh or paraffinised)	