MEDIPOL
İSTANBUL MEDİPOL UNIVERSITY
GENETIC DISEASES ASSESSMENT CENTER

# GYNECOLOGY & UROLOGY TEST LIST

Please Tick the Related Box
Fill in the Anamnesis & Consent Form on the Back Page

| CYTOGENETIC/MOLECULAR CYTOGENETIC TESTS | SAMPLE TYPE |
|---|---|
| ☐ Chromosome Analysis | Blood[Heparin] |
| ☐ Chromosome Analysis | Cord Blood[Heparin] |
| ☐ Chromosome Analysis | Abort Material |
| ☐ Chromosome Analysis | Amniotic Fluid |
| ☐ Chromosome Analysis | CVS |
| ☐ Rapid Aneuploidy FISH (Chromosomes 13, 18, 21, X and Y) | A.F., CVS, Abort, Blood[HEP] |
| ☐ Array CGH | A.F., CVS, Abort, Blood[EDTA] |
| ☐ Maternal Contamination | Blood[EDTA] |
| » In order to exclude maternal contamination, maternal blood should be sent along with the prenatal sample. | |
| **PRENATAL MOLECULAR DIAGNOSTIC TESTS** | **SAMPLE TYPE** |
| ☐ Achondroplasia - FGFR3 (G1138A/C) Mutation Analysis | A.F., CVS, Cord Blood[EDTA] |
| ☐ Beta Thalassemia (Known Mutation) | A.F., CVS, Cord Blood[EDTA] |
| ☐ DMD/BMD Deletion /Duplication Analysis | A.F., CVS, Cord Blood[EDTA] |
| ☐ Phenylketonuria Genetic Analysis (Known Mutation) | A.F., CVS, Cord Blood[EDTA] |
| ☐ Cystic Fibrosis Full Gene Sequence Analysis | A.F., CVS, Cord Blood[EDTA] |
| ☐ SMA Deletion Analysis (Known Mutation) | A.F., CVS, Cord Blood[EDTA] |
| ☐ Array CGH | A.F., CVS, Cord Blood[EDTA] |
| ☐ Maternal Contamination | Blood[EDTA] |
| ☐ ..... (Known Mutation Analysis) | A.F., CVS, Cord Blood[EDTA] |
| » In cases where there is no index in molecular tests for prenatal diagnosis or there is no index case, carrier tests of parents must be completed. | |
| **HABITUAL ABORTION PANEL** | **SAMPLE TYPE** |
| ☐ Chromosome Analysis | Abort Material |
| ☐ Chromosome Analysis (Spouses) | Blood[Heparin] |
| THROMBOPHIA PANEL (FEMALE) | |
| ☐ Factor V Leiden G1691A Mutation Analysis | Blood[EDTA] |
| ☐ Prothrombin G20210A Mutation Analysis | Blood[EDTA] |
| ☐ MTHFR C677T Mutation Analysis | Blood[EDTA] |
| ☐ MTHFR A1298C Mutation Analysis | Blood[EDTA] |
| ☐ PAI 4G/5G Mutation Analysis | Blood[EDTA] |
| ☐ Factor V H1299R Mutation Analysis | Blood[EDTA] |
| SUBCLINIC INFECTIONS (FEMALE) | |
| ☐ Chlamydia Trachomatis DNA PCR | Urine, Swap, Thin Prep |
| ☐ CMV DNA PCR Quantitative | A.F., CSF, Urine, Kan[EDTA] |
| ☐ Mycoplasma Genitalium DNA PCR | Urine, Swap, Thin Prep |
| ☐ Ureaplasma Urealyticum DNA PCR | Urine, Swap, Thin Prep |
| URETHRITE PANEL (MALE) | |
| ☐ Chlamydia Trachomatis DNA PCR | Urine, Swap, Thin Prep |
| ☐ Mycoplasma Genithalium DNA PCR | Urine, Swap, Thin Prep |
| ☐ Neisseria Gonorrhoeae DNA PCR | Urine, Swap, Thin Prep |
| ☐ Ureaplasma Urealyticum DNA PCR | Urine, Swap, Thin Prep |
| ARRAY CGH PANEL | |
| ☐ Chromosome Analysis | Abort Material |
| ☐ Array CGH | Abort Material |
| ☐ Maternal Contamination | Blood[EDTA] |
| **MOLECULAR MICROBIOLOGY TESTS** | **SAMPLE TYPE** |
| ☐ Chlamydia Trachomatis DNA PCR | Urine, Swap, Thin Prep |
| ☐ CMV DNA PCR Quantitative | A.F., CSF, Urine, Blood[EDTA] |
| ☐ HBV DNA PCR Quantitative | Serum |
| ☐ HCV RNA PCR KQuantitative | Serum |
| ☐ HHV-6 DNA PCR | Serum, CSF, Blood[EDTA] |
| ☐ HIV 1 RNA PCR Quantitative | Serum, Blood[EDTA] |
| ☐ HPV DNA PCR | Swap, Thin Prep, Biopsy |
| ☐ HPV DNA Genotyping (>100 Genotypes) | Swap, Thin Prep, Biopsy |
| ☐ HSV Type 1 DNA PCR | CSF, Serum, Swap, Blood[EDTA] |
| ☐ HSV Type 2 DNA PCR | CSF, Serum, Swap, Blood[EDTA] |
| ☐ Mycoplasma Genitalium DNA PCR | Urine, Swap, Thin Prep |
| ☐ Mycoplasma Hominis DNA PCR | Urine, Swap, Thin Prep |
| ☐ Neisseria Gonorrhoeae DNA PCR | Urine, Swap, Thin Prep |
| ☐ Rubella RNA PCR | Amniotic Fluid, Blood[EDTA] |
| ☐ TBC DNA PCR | Urine, BAL, Sputum, CSF |
| ☐ Toxoplasma Gondii DNA PCR | A.F., CSF, Blood[EDTA] |
| ☐ Trichomonas Vaginalis DNA PCR | Swap, Thin Prep |

| MOLECULAR MICROBIOLOGY TESTS | SAMPLE TYPE |
|---|---|
| ☐ Ureaplasma Parvum DNA PCR | Swap, Urogenital Discharge, Biopsy |
| ☐ Ureaplasma Urealyticum DNA PCR | Swap, Urogenital Discharge, Biopsy |
| ☐ Varicella Zoster Virus (VZV) DNA PCR | A.F., BOS, Serum, Blood[EDTA] |
| **STD PANEL (There must be at least 2 dry swap samples upon request.)** | **SAMPLE TYPE** |
| ☐ Chlamydia Trachomatis DNA PCR | Swap, Urogenital Discharge, Biopsy |
| ☐ Gardnerella Vaginalis DNA PCR | Swap, Urogenital Discharge, Biopsy |
| ☐ HSV Type 1, 2 DNA PCR | Swap, Urogenital Discharge, Biopsy |
| ☐ Mycoplasma Genitalium DNA PCR | Swap, Urogenital Discharge, Biopsy |
| ☐ Mycoplasma Hominis DNA PCR | Swap, Urogenital Discharge, Biopsy |
| ☐ Neisseria Gonorrhoeae DNA PCR | Swap, Urogenital Discharge, Biopsy |
| ☐ Ureaplasma Parvum DNA PCR | Swap, Urogenital Discharge, Biopsy |
| ☐ Ureaplasma Urealyticum DNA PCR | Swap, Urogenital Discharge, Biopsy |
| ☐ Trichomonas Vaginalis DNA PCR | Swap, Urogenital Discharge, Biopsy |
| **INFERTILITY PANEL** | **SAMPLE TYPE** |
| ☐ Chromosome Analysis (Male & Female) | Blood[Heparin] |
| ☐ Y Chromosome Microdeletion Analysis (Male) | Blood[EDTA] |
| ☐ Cystic Fibrosis Gene Analysis (Including 5T Variant) | Blood[EDTA] |
| **CARDIOVASCULAR RISK TESTS** | **SAMPLE TYPE** |
| ☐ Factor V Leiden G1691A Mutation Analysis | Blood[EDTA] |
| ☐ Prothrombin G20210A Mutation Analysis | Blood[EDTA] |
| ☐ MTHFR C677T Mutation Analysis | Blood[EDTA] |
| ☐ MTHFR A1298C Mutation Analysis | Blood[EDTA] |
| ☐ PAI 4G/5G Mutation Analysis | Blood[EDTA] |
| ☐ Factor XIII V34L Mutation Analysis | Blood[EDTA] |
| ☐ Beta Fibrinogen-455 G>A Mutation Analysis | Blood[EDTA] |
| ☐ APO B R3500Q Mutation Analysis | Blood[EDTA] |
| ☐ Factor V H1299R Mutation Analysis | Blood[EDTA] |
| ☐ APO-E Genotyping (E2, E3, E4) | Blood[EDTA] |
| ☐ ACE I/D Genotyping | Blood[EDTA] |
| ☐ HPA-1 a/b Genotyping | Blood[EDTA] |
| **MOLECULAR GENETIC TESTS** | **SAMPLE TYPE** |
| ☐ Androgen Receptor-AR Whole Gene Sequence Analysis | Blood[EDTA] |
| ☐ Fragile X Syndrome-FMR1 CGG Repeat Genetic Analysis | Blood[EDTA] |
| ☐ Spinal Muscular Atrophy Carrier Test (SMN1 Deletion/Duplication Analysis) | Blood[EDTA] |
| ☐ Male Infertility Panel - 63 Genes (NGS) | Blood[EDTA] |
| ☐ Female Infertility Panel - 44 Genes (NGS) | Blood[EDTA] |
| ☐ Cystic Fibrosis 5T Variant Analysis - CFTR [IVS8 (5T)] | Blood[EDTA] |
| ☐ Cystic Fibrosis Full Gene Sequence Analysis | Blood[EDTA] |
| ☐ IVF DNA Profile Analysis (ID Required) | Blood[EDTA] |
| ☐ Thyroid Hormone Resistance-THRB Full Gene Sequence Analysis | Blood[EDTA] |
| ☐ Y Chromosome Microdeletion Analysis (25 Sub-Loci) | Blood[EDTA] |
| ☐ BRCA1, BRCA2 Full Gene Sequence Analysis | Blood[EDTA] |
| ☐ DMD/BMD Deletion/Duplication Analysis | Blood[EDTA] |
| ☐ Genetic Carrier Test | Blood[EDTA] |
| ☐ Cancer Genetics Panel - 94 Genes (NGS) | Blood[EDTA] |
| ☐ Breast-Ovarian Cancer Comprehensive Panel - 23 Genes (NGS) | Blood[EDTA] |
| ☐ Non Invasive Prenatal Test (☐ Basic Panel / ☐ Comprehensive Panel) | Special Tube |
| ☐ Sequencing Panel for Premature Ovarian Failure-12 Gene (NGS) | Blood[EDTA] |
| ☐ Clinical Exome Sequence Analysis | Blood[EDTA] |
| ☐ Whole Exome Sequence Analysis (WES) (Please request a detailed consent form) | Blood[EDTA] |
| ☐ Trio Exome Sequence Analysis (TRIO WES) (Please request a detailed consent form) | Blood[EDTA] |

» In cytogenetic studies, due to the nature of the test, living cells must be obtained, and the samples must be delivered to our laboratory maximum within 48 hours under appropriate storage and transport conditions.
» The rate of non-growth in tissue cultures prepared from discharge material is between 10-60%. Changeable. (Ref: European Journal of Human Genetics (2001) 9, 539-547)
» For abortion samples, sending the entire sample increases the risk of possible contamination of the sample with the infectious factor, so it is more appropriate to send a piece of 1 cm3 depth.
»The abortion sample taken should be placed in transport medium or sterile container-saline, it should never be treated with formol/alcohol.

The tests performed in our laboratory are controlled by external quality control programs [QCMD, MOTAKK, CEQAS, EMQN] in the fields of molecular microbiology, molecular cytogenetics and molecular genetics.

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