

OBSTETRICS & GYNECOLOGY TEST MENU

#	Test	Indication	Sample	Turnaround Time
RECURRENT PREGNANCY LOSS				
1	Karyotype - couple	Chromosome analysis in male & female, for detection of chromosomal numerical and structural aneuploidies	WB (Heparinized)	2-3 weeks
2	Array-CGH - couple	(Array-based Comparative Genomic Hybridization) male & female & product of conception to detect any loss or gain in chromosomes that could lead to RPL	WB (EDTA)	2-3 weeks
3	Thrombophilia panel screen - Female	Study of 12 polymorphisms and mutations in female could increase thrombosis tendency in mother through pregnancy, includes FII Prothrombin, FV Leiden, FV 1299, FV Cambridge, FV Y1702C, MTHFR 677, MTHFR 1298, MTR 2756, MTRR 66, FXIII Val34Leu, Beta-Fibrinogen 455 G>A and PAI-1 4G/5G mutations.	WB (EDTA)	3-5 Days
4	Y Chromosome Microdeletions - Male	Microdeletions of the Y chromosome as cause of spermatogenetic failure resulting in male infertility. The molecular detection of deletions has become an important diagnostic test in male infertility studies. Four Azoospermia factors (AZFa, AZFb and AZFc) have been mapped to Yq11, of which AZFc is the most frequent region involved in deletions.	WB (EDTA)	3-5 Days
5	HLA-G - Female	Analysis of polymorphisms -3' UTR 14bp ins/del and -5' UTR-725 C-G.	WB (EDTA)	3-4 weeks
6	Karyotype, product of conception	Chromosome analysis of product of conception (maximum 48hours after missing the fetus) with high-resolution G banding for detection of chromosomal numerical and structural aneuploidies in fetus	POC	2-3 weeks
7	BAC array CGH, product of conception	Combined with maternal cell contamination ruled out to detect chromosomal loss or gain less than 1 MB in fetus before 20 weeks	POC	2-3 weeks
8	Oligonucleotide array CGH, product of conception	To detect chromosomal loss or gain less than 1 MB in fetus after 20 weeks	POC	2-3 weeks
MALE INFERTILITY				
1	Karyotype	Chromosome analysis of male partner. A karyotype is recommended in all men with a total motile sperm count below 5 to 10 million who are thought to have non-obstructive azoospermia.	WB (Heparinized)	2-3 weeks
2	Y Chromosome Microdeletions	molecular detection of microdeletions of the Y chromosome as cause of spermatogenetic failure resulting in male infertility.	WB (EDTA)	3-5 Days
3	FSHR and FSHB Genes study	Analysis of the polymorphisms Ser680Asn of the gene FSHR (FSH Receptor) and - 211 TT (g.4790 G>T) of the gene FSHB, to evaluate the possible treatment of male infertility with FSH hormone	WB (EDTA)	3-4 weeks
4	Male Infertility NGS Panel	Male infertility NGS panel that analyzes 50 genes associated with male infertility. It reveals genetic factors that are associated with some of the most common reproductive conditions in men.	WB (EDTA)	6 weeks

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5	STI Panel	STIs can affect sperm health, including motility, concentration, and morphology, potentially leading to infertility.	Urine or urethral sample	3-5 Days
6	Cystic Fibrosis (CF)	CF testing is indicated for male infertility, especially if they have Congenital Absence of the Vas Deferens (CBAVD). CFTR gene common mutations test is the first step.	WB (EDTA)	3-5 Days
7	Sperm DNA Fragmentation Test, Flow Cytometric	Flow Cytometric SDF (FC-SDF) assay has gained clinical importance compared to commonly used techniques due to its sensitivity (due to analysis of large number of sperm cells) which enables quantitative and subjective test results. TUNEL assay quantifies the incorporation of fluoresceinated dUTP antibody into single- and double-strand DNA breaks.	SF special tube	2 weeks
8	Sperm Precursor Cell Analysis, Flow Cytometric	Reliable prediction before TESE operation to detection of spermatogenic serial cells in the haploid structure that have completed meiosis process in the testis biopsy material or seminal plasma	SF special tube	2 weeks
FEMALE INFERTILITY				
1	Karyotype	Chromosome analysis of female partner	PB (Heparinized)	2-3 weeks
2	Karyotype for Mosaicism	Chromosomal analysis on 50 cells culture to detect mosaicism especially in suspected mosaic Turner syndrome	PB (Heparinized)	2-3 weeks
3	Female Infertility NGS Panel	A comprehensive next generation sequencing (NGS) panel that analyzes 70+ genes associated with female infertility, including primary ovarian insufficiency, Ovarian Dysfunction, Ovarian Dysgenesis, polycystic ovary syndrome, Preimplantation embryonic lethality, errors in folliculogenesis and oocyte maturation defects, ovarian hyperstimulation syndrome, and recurrent pregnancy loss	PB (EDTA)	6 weeks
4	Endometrial Microbiome Analysis	Assessment of the endometrial microbiome to improve the reproductive outcome of infertile patients. Endometrial Microbiome Analysis test provides a complete view of the endometrial bacterial composition, reporting the 10 most represented bacteria in the endometrium, as well as identifying the 8 most common pathogens causing chronic endometritis (CE). Endometrial Microbiome Analysis test can determine whether the uterine microbial environment is optimal for embryo implantation. Depending on the results, it recommends embryo transfer or antibiotic and probiotic treatment, if needed, to restore an optimal microbiome.	Endometrial Biopsy	3-4 weeks
5	Primary Ovarian Insufficiency/Dysfunction NGS	A comprehensive next-generation sequencing (NGS) panel that analyzes 50+ genes associated with primary ovarian insufficiency and Ovarian Dysfunction	PB (EDTA)	6 weeks
6	Ovarian Dysgenesis NGS panel	A comprehensive next-generation sequencing (NGS) panel that analyzes 11 genes associated with Ovarian Dysgenesis	PB (EDTA)	6 weeks
7	Polycystic Ovary Syndrome Genes	A test which analyze the genes CAPN10 and IRS-1, related to polycystic ovary syndrome	PB (EDTA)	4weeks
8	Ovarian Hyperstimulation Syndrome NGS	A panel which investigate 5 genes related to ovary stimulation response: CYP19, ESR1, ESR2, FSHR, IRS1	PB (EDTA)	4weeks

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9	FSHR (FSH Receptor) ovary stimulation response	Analysis of the polymorphisms Asn680Ser and Thr307Ala of the FSHR - FSH Receptor to modulated in individually the administration of FSH to improve the efficacy of therapy	PB (EDTA)	4 weeks
10	IRS1 Gene - Polycystic Ovary, Metformin treatment response	Evaluation of the possible response to a Metformin treatment in women affected by Polycystic Ovary Syndrome (PCOS) or diabetes mellitus type 2, through the analysis of the polymorphism Gly972Arg of the gene IRS-1 (Insulin Receptor Substrate-1	PB (EDTA)	4weeks
11	LHR (LH receptor)	Analysis of the mutation R554X of the gene LHR	PB (EDTA)	4 weeks
12	LHB (Luteinizing hormone subunit beta)	Analysis of the mutations W8R and I15T of the gene LHR	PB (EDTA)	4 weeks
13	ER (Endometrial Receptivity Map)	ER Map® is a personalised test to diagnose the receptivity status of the endometrium during the window of Implantation (WOI).	Endometrial Biopsy - Special Tube	4 weeks
14	STI Panel	Most cases of tubal factor infertility are attributable to untreated sexually transmitted diseases that ascend along the reproductive tract. The panel includes NAAT (Nucleic Acid Amplification Test), a highly sensitive and specific method for detecting bacterial DNA or RNA, for chlamydia, gonorrhea and mycoplasma.	Urine or vaginal sample	3-5 Days

PREMATURE MENOPAUSE

1	Primary Ovarian Insufficiency/Dysfunction NGS	A comprehensive next-generation sequencing (NGS) panel that analyzes 50+ genes associated with primary ovarian insufficiency and Ovarian Dysfunction	WB (EDTA)	6 weeks
2	Fragile X Syndrome - Cyto	Cytogenetic study for Fragile X Syndrome. Fragile X syndrome (FXS) is the leading inherited form of intellectual disability and autism spectrum disorder, and patients can present with severe behavioural alterations.	WB (Heparinized)	2-3 weeks
3	Fragile X Syndrome - Mol	Molecular study, Trinucleotide Repeat expansion	WB (EDTA)	2-3 weeks

PRIMARY AMENORRHEA

1	Karyotype	To reveal aneuploidy, such as Turner syndrome (45, X), or mosaicism, where different cell lines have different chromosome numbers.	WB (Heparinized)	2-3 weeks
2	Karyotype for Mosaicism	Chromosomal analysis on 50 cells culture to detect mosaicism especially in suspected mosaic Turner syndrome	WB (Heparinized)	2-3 weeks
3	Array-CGH	Characterization of an unbalanced X-autosome translocation. Array-CGH was found to be a very useful tool in the identification of candidate genes in conditions associated with primary amenorrhea and ovarian failure.	WB (EDTA)	3-4 weeks
4	Primary Ovarian Insufficiency/Dysfunction NGS	A comprehensive next-generation sequencing (NGS) panel that analyzes 50+ genes associated with primary ovarian insufficiency and dysfunction	WB (EDTA)	6 weeks

#	Test	Indication	Sample	Turnaround Time
PRENATAL SCREENING				
1	NIPT-5	Non-invasive prenatal testing, Screening for chromosomal abnormality in chromosomes 13,18,21,X, and Y plus sex determination (optional) after 10 weeks of gestational age.	WB-Maternal	10-14 days
2	NIPT Karyo	1-Screening for all chromosomal aneuploidies and deletion-duplications in all chromosomes. 2-Fetal sex determination	WB-Maternal	2-3 weeks
3	NIPT Rh	Early non-invasive determination of fetal Rh(D) factor genotyping through analysis of free fetal DNA isolated from maternal blood beyond 10 weeks	WB-Maternal	2-3 weeks
4	First Trimester Maternal Serum Screen (NT, hCG, PAPP-A)	This test requires a nuchal translucency (NT) measurement performed by a certified ultrasonographer. This test does not screen for Open Neural Tube Defect (ONTD) and is only used to screen for fetal risk of Down syndrome (trisomy 21) and trisomy 18. Specimen must be drawn in the first trimester between 11 weeks, 0 days and 13 weeks, 6 days. (Crown-Rump length (CRL) must be between 4.2-8.5 cm).	Maternal serum	7-10 days
5	Triple Marker Test (AFP, hCG, & Estriol)	Specimen must be drawn between 14 weeks, 0 days and 24 weeks, 6 days gestation. This test is used to screen for fetal risk of Down syndrome (trisomy 21), trisomy 18, and Open Neural Tube Defect (ONTD, spina bifida).	Maternal serum	7-10 days
6	Quad Marker Test (AFP, hCG, Estriol, Inhibin A	Specimen must be drawn between 14 weeks, 0 days and 24 weeks, 6 days gestation. This test is used to screen for fetal risk of Down syndrome (trisomy 21), trisomy 18, and Open Neural Tube Defect (ONTD, spina bifida).	Maternal serum	7-10 days
AMBIGUOUS GENITALIA				
1	Karyotype	A 46,XY karyotype with ambiguous genitalia suggests disorders of sex development (DSDs) and in 46,XX individuals, congenital adrenal hyperplasia (CAH) is a common cause.	WB (Heparinized)	2-3 weeks
2	Congenital Adrenal Hyperplasia (CAH)	CAH causes the adrenal glands to produce excessive androgens, leading to virilization of the external genitalia in females.	WB (EDTA)	7-10 days
3	Sexual Developmental Disorders NGS Panel	Clinical suspicion of an intersex condition, presenting with ambiguous genitalia, abnormal gonadal structure or function, or related symptoms like delayed or abnormal puberty.	WB (EDTA)	6 weeks
PRENATAL DIAGNOSIS (PND)				
1	Karyotype, Amniotic fluid or CVS	Prenatal Diagnosis of numerical and structural chromosomal aneuploidy in fetus	WB (Heparinized)	2-3 weeks
2	Carrier gene analysis	in parents for prenatal diagnosis of any genetic disease, best time is before pregnancy of maximum beginning of it	WB (EDTA)	4 weeks
3	β- thalassemia, prenatal diagnosis	Prenatal Diagnosis for β Thalassemia through CVS at 12-14 weeks or amniocentesis in 14-17 weeks	CVS-AF	2-3 weeks

#	Test	Indication	Sample	Turnaround Time
4	Prenatal diagnosis After mutations analysis in the family	Prenatal Diagnosis for all of these disease and other genetic conditions in the family through CVS at 12-14 weeks or amniocentesis at 14-17 weeks • α- thalassemia • Sickle cell anemia • Cystic Fibrosis, • Osteogenesis Imperfecta • Hemophilia A or B • Duchenne & Becker Muscular Dystrophy • Spinal Muscular Atrophy	CVS - AF	2-3 weeks
INFECTIOUS DISEASES				
1	Cytomegalovirus (CMV) Detection	Detection of CMV virus in prenatal period in amniotic fluid or cervicovaginal smear by real time PCR	WB (EDTA)	Next day
2	STI Panel	NAAT (Nucleic Acid Amplification Test), a highly sensitive and specific method for detecting bacterial DNA or RNA, for chlamydia trachomatis, Neisseria gonorrhea and mycoplasma genitalium.	Urine or vaginal sample	3-5 Days
HEREDITARY BREAST & OVARIAN CANCER				
1	BRCA1 & BRCA2 genes	For hereditary breast & ovarian cancer screening, NGS		3-4 weeks
2	Breast cancer susceptibility panel – 21 genes	21 genes study (BRCA1 & BRCA2, CHECK2, PALB2, BRIP1, MLH1,...), NGS		6 weeks
3	HER2 gene presentation	For treatment with herceptine, FISH method	WB (EDTA) / Bone Marrow / Paraffin Block	2-3 week
PREIMPLANTATION GENETIC TESTS (PGT)				
1	PGT-A – NGS	Chromosomal Aneuploidy Screening in embryos with NGS method	Trophectoderm /Blastomere in special tube	10 Days
2	PGT-SR – NGS	Study of embryos for chromosomal structural anomalies by NGS method	Same as above	10 Days
CERVICAL CANCER SCREEN				
1	HPV Genotyping	Cervical cancer screening guidelines advise HPV testing in women ages 30 to 64 years as co-testing with cervical cytology or as a reflex test when the Pap test result is atypical squamous cells of undetermined significance (ASC-US).	Cervical smear	Next day

#	Test	Indication	Sample	Turnaround Time
2	Liquid pap cytology	Liquid-based cytology (LBC) is a method of preparing cervical samples for cytological examination. Unlike the conventional 'smear' preparation, it involves making a suspension of cells from the sample and this is used to produce a thin layer of cells on a slide.	Cervical smear	7-10 Days
3	Conventional slide pap smear		Cervical smear	Next day
MISCELLANEOUS				
1	Whole Exome Sequencing - solo	Recommended for complex and undiagnosed cases suspicion of genetic causes. It Diagnose more than 7000 rare monogenic disease plus chromosomal microdeletion, duplications and mitochondrial disease.	Dried Blood Spot, WB (EDTA), Tissue, AF	6 weeks
2	Whole Exome Sequencing- Double	For couples with history of passed offspring suspected to have genetic disease to detect carrier status	Dried Blood Spot, WB (EDTA), Tissue, AF	6 weeks
3	Congenital Adrenal Hyperplasia common mutations of CYP21A2	As first step of genetic study for diagnosis of Congenital adrenal hyperplasia (CAH) as the most common cause of ambiguous genitalia	WB (EDTA)	3-5 days
4	CFTR gene common mutation (18 mutations ΔF508 included)	CYSTIC FIBROSIS (CF) MULTIPLEX real time PCR test a first genetic study for CF diagnosis	WB (EDTA)	3-5 days
5	MEFV gene common mutations (20 mutation)	Familial Mediterranean Fever (FMF) MULTIPLEX real time PCR test for detection of 20 common mutations in this gene that is covering 99.2% mutation rate of FMF in the Anatolian, Middle East countries and many other countries.	WB (EDTA)	3-5 days
6	Sex Determination, SRY Gene	AMXY gene on X and Y chromosome and SRY gene on Y chromosome are studies through PCR for determining the sex of individual or fetus	WB (EDTA), AF, CVS	2-3 weeks
7	Newborn Screening Test (from Austria)	Comprehensive screening programs including biochemical analysis of over 48 Inborn Errors of Metabolism and other disorders. Best time is in first week after birth.	Dried Blood Spot	10 days
8	Umbilical Cord Blood Banking	Storage of cord blood that is rich in hematopoietic stem cells, similar to those found in bone marrow. Cord blood can be used for transplantation as an alternative to bone marrow.	Call us	