CLINICA SANTE
SPECIALIȘTI ÎN ANALIZE MEDICALE

FISA PACIENTULUI

Va rugam insistent sa completati toate campurile!

ID Pacient_____

DATE PROBA: Data recoltarii: ____ /____ /_____ Data primirii: ___ /____ /_____
SPITAL /CLINICA: _____
Telefon: _____Email: _____
MEDIC:_____
Telefon: _____Email: _____
DATE PACIENT: Nume : _____
Prenume:_____
Data nasterii: ___ /___ /_____ Sex: M ☐ F ☐
CNP:_____
Adresa:_____
Telefon:_____Email:_____
Nume partener: _____
Data ultimei menstruatii (DUM):___/___/_____ ☐ S. dubla ☐ S. Multipla
Varsta sarcinii (DUM) : _____ Varsta sarcinii (ECO) : _____

☐Lichid amniotic
☐Vilozitati coriale
☐Sange periferic
☐Sange fetal
☐Ser
☐Urina
☐Lichid seminal
☐Produs de avort
☐Maduva osoasa
☐Celule blastice periferice
☐Tampon endocervical, vaginal, uretral
☐Altele

INDICATII (OBLIGATORIU)

☐ **Varsta materna:** _____ ☐ **Boli genetice in familie:** _____
☐ **Translucenta nucala_____mm_____** ☐ **Probleme de fertilitate_____**
☐ **Alte constatari din Eco_____** ☐ **Fenotip anormal_____**
☐ **Risc crescut dupa controlul biochimic prenatal:** _____ ☐ **Avorturi repetate_____**
☐ **Purtator al unei anomalii cromozomiale/boli monogenice:** _____ ☐ **Investigarea identitatii ADN** _____
☐ **Control imunologic pozitiv_____** ☐ **Altele:** _____
Explicatii: _____

☐ **CONSULT GENETIC**

TEST NON INVAZIV DIN ADN FETAL LIBER CIRCULANT IN SANGE MATERN (cffDNA) (fisa speciala)

☐ Harmony – Roche trisomiile 13,18,21 ☐ Sex fetal si aneuploidii X,Y
☐ Veracity-NIPD Genetics trisomiile 13,18,21 ☐ Sex fetal, aneuploidii X, Y si panel Microdeletii
☐ Genome-Wide NIPT – Genoma - PrenatalSafe Karyo ☐ PrenatalSafe Karyo + (plus 9 microdeletii)

NOU **WHOLE GENOME ANALYSIS – SNP microarray**

☐ Cariotip Molecular (SNP microarray) Lichid amniotic/CVS (Vilozitati coriale): include si QF-PCR (13,18,21,X,Y)
☐ Cariotip Molecular (SNP microarray) Sange periferic

CITOGENETICA CONVENTIONALA PRENATAL/POSTNATAL

☐ Alfa fetoproteina din lichid amniotic
☐ Cariotip clasic Lichid amniotic/ CVS (Vilozitati coriale)
☐ Cariotip clasic Lichid amniotic/ CVS (Vilozitati coriale) plus QF-PCR (13,18,21,X,Y)
☐ ***NOU*** Cariotip clasic Lichid amniotic / Vilozitati coriale (CVS) si Test Rapid Extins MLPA care include: aneuploidii 13,18,21 si analiza 20 sindroame microdeletii
☐ Cariotip produs de conceptie (avort) ☐ Cariotip sange fetal
☐ Cariotip sange periferic High Resolution
☐ Cariotip maduva osoasa ☐ Cariotip blastice periferice
☐ FISH cu probe specifice prenatal/postnatal Specificati:
☐ Fragmentare ADN, sperma (metoda TUNEL)/morfologie sperma

MOLECULARE

- ☐ Acondroplazie gena *FGFR3* mutatiile G1138A si G1138C
- ☐ Atrofia musculara spinala (SMA) (exonii 7 si 8 ai genelor *SMN1* si *SMN2*) certificat IVD (In vitro diagnostic)
- ☐ ***NOU*** Boala cardiaca congenitala (MLPA)
- ☐ Charcot-Marie-Tooth gena *PMP 22* ☐ Charcot-Marie-Tooth gena *GJB1* (X-linkat)
- ☐ ***NOU*** Deletii ale genei *SHOX*
- ☐ Detectia genei *SRY*
- ☐ ***NOU*** Disomie uniparentala cromozomii 7/14
- ☐ Fibroza chistica gena *CFTR* mutatia DF508 ☐ Fibroza chistica gena *CFTR* cele mai comune 39 mutatii
- ☐ Fragile X (FRAXA)
- ☐ Hipocondroplazie gena *FGFR3* mutatiile C1620A si C1620G
- ☐ Microdeletii cromozomul Y (*AZF*)
- ☐ Prader Willi/Angelman metilare (MS-MLPA)
- ☐ Retard mental X-linkat
- ☐ Rinichi polichistic gena *PKD1* si *PKD2*
- ☐ Screening 20 sindroame microdeletii prenatal/ postnatal
- ☐ Screening microdeletii/microduplicatii subtelomerice ale cromozomilor
- ☐ ***NOU*** Sindrom DiGeorge (MLPA)
- ☐ Sindrom Duchenne gena *DMD* (MLPA)
- ☐ ***NOU*** Sindrom Russel-Sylver/Beckwith-Wiedemann (MLPA)
- ☐ Surditate ereditara mutatia 35delG gena *GJB2* ☐ Surditate ereditara mutatia W24X gena *GJB2*
- ☐ Surditate ereditara genele *GJB2*, *GJB3*, *GJB6* cele mai comune mutatii
- ☐ Talasemia gena *HBB* cele mai commune 22 mutatii
- ☐ Test Rapid QF-PCR: aneuploidii 13,18,21,X,Y
- ☐ ***NOU*** Test Rapid Extins MLPA: aneuploidii 13,18,21 si analiza 20 sindroame microdeletii

NOU !!! **NGS –SECVENTIEREA PE PANELURI DE GENE PENTRU BOLI GENETICE**

- ☐ Secventierea intregului exom (consult genetic inclus)
- ☐ Test screening myBRCA-gene BRCA1 si BRCA2 (consult genetic inclus)
- ☐ Test screening myBRCAHiRisk- 26 gene (consult genetic inclus)

TROMBOFILIE

- ☐ Factorul II (protrombina) ☐ Factorul V Leiden ☐ Factorul V R2
- ☐ MTHFR 677 si 1298 ☐ Factorul XIII Val34Leu
- ☐ PAI1 - 1 4G/5G ☐ Gena ACE polimorfism I/D

VIRUSOLOGIE SI BACTERIOLOGIE MOLECULARA

- ☐ Detectie ADN de CMV (Citomegalovirus) / ☐ EBV (Epstein-Barr Virus) / ☐ VSH 1 si 2 (Herpes Simplex Virus) / ☐ VZV (Varicella Zoster Virus) – 18 saptamani de sarcina
- ☐ Detectie ADN de Toxoplasma gondii – 18 saptamani de sarcina ☐ Detectie ARN de Rubella Virus

NOU !!! **Panel boli cu transmitere sexuala (11 patogeni)**

- ☐ Detectie ADN de: Chlamydia trachomatis, Haemophilus ducreyi, Herpes simplex viruses (HSV-1/HSV-2), Mycoplasma genitalium, Mycoplasma hominis, Neisseria gonorrhoeae, Treponema pallidum, Trichomonas vaginalis and Ureaplasma (urealyticum/parvum)

PACHETE

- ☐ Cariotip Sange High Resolution + Fibroza chistica gena CFTR cele mai comune 39 mutatii + Microdeletii cromozomul Y (AZF)
- ☐ Cariotip Sange High Resolution + Fraxile X (FRAXA) + Panel trombofilie (FactorII+FactorV Leiden+MTHFR 677 si 1298)
- ☐ Factorul II + Factorul V Leiden + Factorul V R2 + *MTHFR* 677 si 1298 + *PAI1*- 4G/5G + Factorul XIII Val34Leu + *ACE* I/D
- ☐ Dublu Test (PAPP-A, free-beta HCG) + Fibroza chistica gena CFTR mutatia DF508 + Surditate ereditara gena GJB2 mutatia 35delG
- ☐ Dublu Test (PAPP-A, free-beta HCG) + Fibroza chistica gena CFTR mutatia DF508 + Surditate ereditara gena GJB2 mutatia 35delG + Atrofia musculara spinala (SMA) (exonii 7 si 8 ai genelor SMN1 si SMN2)
- ☐ Cariotip produs de avort + Detectie ADN de CMV (Citomegalovirus) + Detectie ADN de Toxoplasma gondii
- ☐ ***NOU*** Cariotip produs de avort + Screening 20 sindroame microdeletii