THE CYPRUS INSTITUTE OF NEUROLOGY & GENETICS
DEPARTMENT OF CYTOGENETICS & GENOMICS
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DEPARTMENT OF CYTOGENETICS & GENOMICS – REFERRAL LETTER

Department Code: 29

Lab no. _____

PATIENT DETAILS

Name:		ID no.:	
Address:		Tel no. home:	
		Tel no. mobile:	
CING no.:	Clinic File no.:	Hospital File no.:	Nationality:
Date of Birth:	Gender: ☐ Male ☐ Female ☐ Fetus		

Partner's name:	Date of Birth:	ID no.:

PATIENT STATUS

Case type: ☐ Outpatient ☐ Inpatient

GESY: ☐ | Government-Non GESY: ☐ | Hospital Card no: | Private-Non GESY: ☐

REFERRING PHYSICIAN/SCIENTIST – STATUS

CING: ☐ | Government (ΟΚΥπΥ): ☐ | Private-GESY: ☐ GESY no: | Private-Non GESY: ☐

Name:			
Address:		Tel no.:	
		Fax no.:	
Hospital/Clinic:		Email:	

The referring physician undertakes and confirms understanding and compliance in respect of the mutual obligations as these are determined under the GDPR.

CLINICAL INFORMATION

Reason for Referral:

Sample type: | Collection date: / / Biopsy date: / / | Time:

LMP: / / | Gestational age in weeks: | Family no.:

Delivered by:	Received by:	Time:	Date: ___/___/___

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TYPE OF SERVICE

CLINICAL CYTOGENETICS – CHROMOSOMAL ANALYSIS of

- ○ 1*. Chorionic Villus Sample (CVS) (Transport Medium)
- ○ 2*. Amniotic Fluid (AF) (20ml)
- ○ 3*. Fetal Blood (FB) (2ml in Sodium or Lithium Heparin)
- ○ 4*. Peripheral Blood (PB) (5ml in Sodium Heparin)
- ○ 5*. Skin Biopsy (SB) (Transport Medium/Normal Sterile Saline)
- ○ 6*. Examination of Products of Conception (POC) (Transport Medium/Normal Sterile Saline)
- ○ 7. Tissue Culture only – (TC) To facilitate other tests
- ○ 8*. Peripheral Blood for couples (PB) (5ml in Sodium Heparin)

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Patient Name:	Lab no.: _____

MOLECULAR CYTOGENETICS

- ○ 20*. Identification/Confirmation/Characterisation of chromosomal abnormality(ies) by Fluorescence In Situ Hybridization (FISH) (F) (5ml PB or 2ml FB in Sodium Heparin, AF, CVS)
- ○ 21*. Diagnosis of the following diseases/syndromes by FISH (F) (5ml PB or 2ml FB in Sodium Heparin, AF, CVS)
  - ☐ Prader-Willi/Angelman (D15S10) ☐ Williams ☐ Miller-Dieker ☐ Cri du chat
  - ☐ Prader-Willi/Angelman (SNPRN) ☐ X-Linked Ichthyosis ☐ 22q11.2 deletion ☐ Kallmann
  - ☐ Smith-Magenis ☐ Retinoblastoma ☐ Wolf-Hirschhorn ☐ Other
- ○ 22. Multiprobe detection centromeric/telomeric by FISH (MP) (5ml PB or 2ml FB in Sodium Heparin, AF, CVS)
- ○ 25. Sperm FISH for chromosome aneuploidies (Semen)
- ○ 26. Sperm DNA Fragmentation (SF) (Semen)
- ○ 27. Sperm Oxidative Stress (OS) (Semen) (Private only)

DNA ANALYSIS

- ○ 60*. Prenatal Diagnosis of Fragile X Syndrome (FR) (AF, CVS, 2ml FB in EDTA)
- ○ 61*. Postnatal Diagnosis of Fragile X Syndrome (FR) (5ml PB in EDTA)
- ○ 62. Investigation of abnormalities/syndromes with MLPA/qRT-PCR (NF) (5ml PB in EDTA)
  - ☐ MLPA ☐ qRT-PCR ☐ UPD ☐ X-inactivation
- ○ 63. DNA Isolation (DNA) (5ml PB or 2ml FB in EDTA, AF, CVS)
- ○ 64. Detection of Y chromosomal material (Y) (5ml PB in EDTA)
- ○ 65*. Screening for Y (AZF) chromosomal microdeletion (PWA) (5ml PB in EDTA)
- ○ 66. Achondroplasia Mutations G1138A and G1138C analysis (AC) (5ml PB or 2ml FB in EDTA, AF, CVS)
- ○ 67. Central Diabetes Insipidus (CDI) (5ml PB in EDTA)
- ○ 68*. Molecular Diagnosis of Prader Willi/Angelman Syndrome (PWA) (5ml PB in EDTA)
- ○ 69*. Rapid prenatal diagnosis of 13, 18, 21, X, Y aneuploidies with QF-PCR (QF) (AF, CVS, FB)
- ○ 70*. Detection of genomic imbalances with high-resolution microarray-CGH (MK) (5ml PB or 2ml FB in EDTA, AF, CVS)
- ○ 71*. Pre-implantation Genetic Testing with microarray-CGH or NGS (PGT) (Biopsied embryo in 2-3μl PBS)

CELL LINES

- ○ 80. Establish Lymphoblastoid Cell line (CL) (10ml PB in Sodium Heparin)
- ○ 81. Establish Fibroblast Cell Line (CL) (Transport Medium)
- ○ 82. Thawing-Freezing-Expansion up to 2T-25 Flasks (CL)
- ○ 83. Maintain Two Vials/Sample in N2/year (CL)

NEXT GENERATION SEQUENCING – (NGS) – Postnatal referrals: 5ml P. Blood in EDTA from patient, and parents as needed.

Please tick the box if diagnostic NGS tests are being requested and fill in the following fields: ☐ Current NGS referral is for diagnostic purposes only

Please state if any other diagnostic tests have been performed for this patient YES / NO

If YES state which tests: _____

If NO state why patient is referred for NGS as a first-tier test: _____

- ○ 110*. Targeted Panel up to 50 genes (TNGS)
  - ☐ Rett/Angelman-like panel
- ○ 111*. Clinical Exome Sequencing (CES) (Single patient analysis) (CNGS)
- ○ 112*. Clinical Exome Sequencing (Trio analysis) (CNGS)
- ○ 113*. *In Silico* Panel from Clinical Exome Sequencing (INGS) (Single patient analysis)
  - ☐ Brain Malformation panel ☐ Syndromic Autism panel
- ○ 114*. *In Silico* Panel from Clinical Exome Sequencing (INGS) (Trio analysis)
  - ☐ Brain Malformation panel ☐ Syndromic Autism panel
- ○ 115*. Whole Exome Sequencing (WES) (ENGS) (Single patient analysis)
- ○ 116*. Whole Exome Sequencing (ENGS) (Trio analysis)
- ○ 117*. Open up the CES data after in silico panel (RNGS)
- ○ 118*. Open up the WES data after in silico panel (RNGS)
- ○ 119*. Sanger DNA sequencing for confirmation of variant detected by NGS/patient (SEQ)

- ○ 99. Research Area (R): _____

Consent

☐ I authorize the Department of Cytogenetics and Genomics to test my (or my child's/my fetus') sample for the designated genetic condition. My signature below constitutes my acknowledgment that the benefits, risks and limitations of this test have been explained to my satisfaction by a qualified health professional. I understand that all test results are treated with confidentiality and that results will only be provided to the referring clinician.

☐ I authorize the department to store my/my child's/fetus' DNA sample, for future reference or use upon authorization.

Patient's/Guardian's Signature: _____ Date: ____/____/____

Clinician's Signature: _____ Date: ____/____/____