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GENETICS AND GENOMICS DIAGNOSTIC LABORATORY
For courier service and/or inquiries, please contact 513-636-4474 • Fax: 513-636-4373
3333 Burnet Avenue, Room NRB 1013, Cincinnati, OH 45229 (For Saturday delivery, please include "Dock 5" on the airbill)
www.cincinnatichildrens.org/cytogenetics • Email: LabGeneticCounselors@cchmc.org

PRENATAL GENETICS REQUISITION

All Information Must Be Completed Before Sample Can Be Processed

PATIENT INFORMATION

Patient Name: _____ , _____ , _____
Last First MI
Date of Birth: ____ / ____ / ____ Gender: ☐ Female ☐ Male

MRN: _____

Address: _____
_____

Fetal Sex: ☐ Male ☐ Female ☐ Ambiguous ☐ Unknown
Based on: _____

ETHNIC/RACIAL BACKGROUND (Choose All)

☐ European American (White)
☐ African-American/black
☐ Native American or Alaskan
☐ Asian
☐ Pacific Islander
☐ Ashkenazi Jewish ancestry
☐ Latinx/Hispanic: (country/region of origin) _____
☐ Other: (country/region of origin) _____

FETAL SAMPLE INFORMATION

SPECIMEN TYPE:

☐ Amniotic Fluid -- Amnioinfusion performed? Yes ☐ No ☐
☐ CVS ☐ Products of Conception (type:_____)
☐ Fetal Peripheral Blood ☐ Fetal Urine
☐ Other _____

SPECIMEN DATE: _____ / _____ / _____ TIME: _____

DRAWN BY: _____

PREGNANCY DATA: (Multiple gestation: Complete separate requisitions for each fetus)

Gestatational age (GA) at sample collection: _____wks _____days

☐ GA by Ultrasound ☐ GA by LMP

G _____ P _____ SAB _____ TAB _____

PARENTAL SAMPLE INFORMATION

☐ **Maternal Sample (REQUIRED)**

☐ Peripheral Blood ☐ Saliva ☐ Other:_____

Specimen Date: _____ / _____ / _____ Time: _____

Drawn By: _____

☐ Paternal Sample Included

Father's name: _____, _____
Last First
Date of Birth: ____ / ____ / ____ Gender: ☐ Male MRN:_____

☐ Peripheral Blood ☐ Saliva ☐ Other:_____

Specimen Date: _____ / _____ / _____ Time: _____

Drawn By: _____

PROVIDER INFORMATION

Ordering Provider (print): _____

Ordering Provider Title: _____

Institution:_____

Address: _____

Phone: (_____) _____ Fax: (_____) _____

Email: _____

_____ Date: _____/ _____/ _____
Ordering Provider Signature (REQUIRED)

Contact Information for questions/results *(if different than ordering provider)*:

Name & Title: _____

Phone: (_____) _____ Fax: (_____) _____

Email: _____

INDICATIONS/DIAGNOSIS/ICD-10 CODE

☐ Abnormal Screening Test Result
Increased risk of: _____
☐ Abnormal fetal ultrasound: _____
_____
☐ Recurrent Pregnancy Loss
☐ Family History: _____
☐ Advanced Maternal Age
☐ Consanguinity (please specify relationship): _____
☐ Other: _____

BILLING INFORMATION

Please call 1-866-450-4198 with billing questions

☐ INSTITUTION BILL

Institution: _____
Address: _____
City/State/Zip: _____
Accounts Payable Contact Name: _____
Phone: _____
Fax: _____
Email: _____

☐ COMMERCIAL INSURANCE

Can only be billed if requested at time of service.
☐ Billing information attached - include a copy of insurance card/face sheet

Cancellation Policy: Tests can only be cancelled if laboratory is notified prior to the initiation of testing.

☐ Patient signed completed ABN

Medical Necessity Regulations: At the government's request, the Molecular Genetics Laboratories would like to remind all physicians that when ordering tests that will be paid under federal health care programs, including Medicare and Medicaid programs, that these programs will pay only for those tests the relevant program deems to be (1) included as covered services, (2) reasonable, (3) medically necessary for the treatment and diagnosis of the patient, and (4) not for screening purposes.

Please Note: We will not bill Medicaid, Medicaid HMO, or Medicare except for the following: CCHMC Patients, CCHMC Providers, or Designated Regional Counties. If you have questions, please call 1-866-450-4198 for complete details.

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Patient Name: _____ **Date of Birth:** _____

PREVIOUS GENETIC TESTING RESULTS

Please provide a copy of test RESULTS for all previous genetic testing performed at a non-CCHMC laboratory. Select all reports included below:

☐ Chromosome/FISH analysis ☐ Microarray ☐ NIPT Screening ☐ Parental Carrier Screening ☐ Single Gene Sequencing ☐ Other: _____

TESTS REQUESTED

TESTING FOR CHROMOSOMAL DISORDERS

Prenatal Reflex Test (See page 5 for additional information):

☐ **Aneuploidy FISH Panel** (13, 18, 21, X and Y) with **Reflex** to:

- – Chromosome Analysis If FISH is ABNORMAL **OR**
- – SNP Microarray on direct amniotic fluid If FISH is Normal

☐ **Parental testing** for abnormal fetal SNP results (VUS, likely pathogenic or pathogenic): **Microarray Family Study**

☐ Test Maternal sample ☐ Test Paternal sample

☐ Prenatal Aneuploidy FISH Panel (FISH for 13, 18, 21, X and Y)

☐ Other FISH testing (please call lab for availability): _____

_____

☐ Chromosome Analysis

☐ Reflex to Microarray if products of conception sample fails to grow for chromosome analysis

☐ SNP Microarray

☐ Reflex to Special Study if microarray is non-diagnostic

-- Culture and send cells to:_____

A completed external lab requisition must be sent with the sample†

☐ Parental testing for abnormal fetal SNP results (VUS, likely pathogenic or pathogenic): **Microarray Family Study**

☐ Test Maternal sample ☐ Test Paternal sample

☐ Microarray Family Study - **Positive Family Member (FM) Information:**

FM Positive Results: _____

FM Name: _____ FM Date of Birth:_____

OTHER TESTING

☐ Maternal Cell Contamination (MCC•): amniotic fluid & CVS samples ***or*** Maternal Engraftment (ME): cord blood

(Maternal sample required for both options)

For **cord blood** samples only:

Baby's Name: _____ Baby's DOB:_____

☐ Do **NOT** include AFP or ACHE testing in order

☐ ACHE (amniotic fluid)

☐ **Special Study** (please call lab prior to ordering)

-- Culture and send cells to:_____

A completed external lab requisition must be sent with the sample†

☐ **Special Study is the priority over microarray OR**

☐ **Microarray is the priority over special study**

☐ Special Study culture and freeze

☐ DNA extraction & storage (**see backup culture policy on page 5) --

Minimum DNA amount: _____

☐ Thaw and Expand previous sample (**Special Study**)

☐ Fragile X (MCC• required)

- Fragile X repeat expansion testing is available for cultured amniocytes, cultured CVS and POC samples. Fragile X repeat expansion testing with methylation analysis is only available for direct amniotic fluid samples.

TARGETED FAMILIAL TESTING FOR KNOWN VARIANTS

☐ **Known mutation analysis**
(Targeted Sanger sequencing)

☐ **Deletion/Duplication analysis by aCGH**
(Targeted Del/Dup - Please contact the lab prior to ordering to confirm availability)

Gene: _____ Familial variant 1 : _____

Familial variant 2 (if applicable): _____

Family members' test reports must be sent with sample.

***Control samples may be needed (see page 5 for additonal information)

Login control samples for DNA Extraction & Storage (Director Discretion - no charge)

SINGLE GENE TESTING

☐ Spinal Muscular Atrophy - SMN1/SMN2 Copy Number Analysis

☐ Alpha (HBA1/2) and Beta (HBB) Globin Gene Locus Analysis (Panel)

☐ HBA1 and HBA2 (α-globin) sequence analysis

☐ HBA1 and HBA2 (α-globin) locus del/dup analysis (HBA1/2 & HBZ)

☐ HBB (β-globin) sequence analysis

☐ HBB (β-globin) locus del/dup analysis (HBB, HBD, HBG1/2, & HBE)

Full Gene Sequencing - *Director approval is required*

Please select from the single genes listed below.

☐ ACADM	☐ DES	☐ IDS	☐ NKX2-5	☐ TAZ
☐ ABCB11	☐ ELANE	☐ IL2RG	☐ NODAL	☐ TBX1
☐ ABCB4	☐ EMD	☐ ITK	☐ OTOF	☐ TBX5
☐ ACTA2	☐ EYA1	☐ JAG1	☐ POLG1	☐ TGFBR1
☐ ADAMTS13	☐ FANCA	☐ KCNJ2	☐ PRF1	☐ TGFBR2
☐ ALDOB	☐ FANCC	☐ LAMP2	☐ PTEN	☐ TITIN
☐ APOB	☐ FANCG	☐ LDB3	☐ RAB27A	☐ TJP2
☐ ATP7B	☐ FASLG	☐ LDLR	☐ SBDS	☐ TNFRSF6
☐ ATP8B1	☐ FBN1	☐ LMNA	☐ SCO2	☐ TNNT2
☐ BAAT	☐ FOXH1	☐ LRBA	☐ SERPINA1	☐ UGT1A1
☐ CASP10	☐ FOXP3	☐ MAGT1	☐ SH2D1A	☐ VCP
☐ CASQ2	☐ GAA	☐ MAP2K1	☐ SKI	☐ VLCAD
☐ CAV3	☐ GAMT	☐ MECP2	☐ SLC22A5	☐ WAS
☐ CD40LG	☐ GATA2	☐ MYBPC3	☐ SLC26A4	☐ XIAP
☐ CDH23	☐ GATM	☐ MYH11	☐ SLC6A8	☐ ZIC3
☐ CFC1	☐ GBA	☐ MYH7	☐ STAT3	
☐ CPT2	☐ GJB2	☐ MYL2	☐ STX11	
☐ CTLA4	☐ GLA	☐ MYL3	☐ STXBP2	
☐ CTNS	☐ HAX1	☐ MYO7A	☐ SURF1	

Please contact the lab regarding sequencing of any other single genes.

INFECTIOUS DISEASE TESTING

Performed by Molecular and Genomic Pathology Services (MGPS) Lab

☐ Cytomegalovirus qualitative PCR

☐ Herpes Simplex Virus 1 and 2 qualitative PCR

☐ Parvovirus qualitative PCR

☐ Toxoplasma gondii qualitative PCR

☐ Other: _____

See page 5 for additional details

†If all requisition forms for recipient lab are not received within 1 week of our sample receipt, the sample will be frozen and stored. Please check with special study recipient lab for additional required materials (such as maternal sample) that must be sent with the proband sample.

•Prenatal samples that require additional culturing for MCC testing will incur an additional fee.

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Patient Name: _____ Date of Birth: _____

GENE PANEL TESTING

☐ Cleft and Craniofacial Gene Panel (288 genes)*

☐ **Reflex to Whole Exome Sequencing**
Whole exome sequencing (WES) orders require a signed WES Consent Form and completion of the WES Test Requisition. Also, inclusion of biological parental samples is strongly encouraged to assist with the analysis of WES and to increase test yield. Please visit our website at www.cincinnatichildrens.org/exome to obtain the required documents. WES testing will NOT be started until all forms are completed and received by the lab.

☐ **Parental Testing** for abnormal fetal results (pathogenic, likely pathogenic or VUS) for the Cleft and Craniofacial Gene Panel: (Family Study)
☐ Test Maternal sample ☐ Test Paternal sample

CARDIOLOGY GENE PANELS

☐ Congenital Heart Diseases Panel (187 genes)*
☐ Heterotaxy Panel (114 genes)*
☐ RASopathy-Noonan Panel (31 genes)*

☐ **Reflex to deletion/duplication of all genes** available on the **cardiology panel selected above***
☐ **Reflex to deletion/duplication of single gene(s)** available on the **cardiology panel selected above*** (specify):_____
☐ **Parental Testing** for abnormal fetal results (pathogenic, likely pathogenic or VUS) for the **cardiology gene panel or del/dup test selected above**: Gene Panel or Targeted del/dup by CGH (Family Study)
☐ Test Maternal sample ☐ Test Paternal sample

*See page 4 for additional gene panel and del/dup information

PRENATAL EXOME TESTING

☐ **Prenatal Exome Sequencing**
☐ Fetus only
☐ Duo (fetus and biological mother)
☐ Trio (fetus and both biological parents)

Preliminary Results
A verbal preliminary result can be provided in 15 days for a provider-defined list of genes (up to 15 genes)

Gene list: _____
_____
_____
_____

Order checklist:
☐ Fetal sample
☐ Maternal sample
☐ Paternal sample and demographic information (page 1)
☐ Preliminary gene list
☐ Detailed clinical information (see below)
☐ Family history/pedigree
☐ Completed consent form

Providing a copy of clinical records including imaging reports (e.g. MRI, ultrasound, echocardiogram) is strongly recommended.

CLINICAL INFORMATION - REQUIRED for Prenatal Exome Sequencing

Please check all that apply and specify in the space provided

Abdomen and Gastrointestinal
☐ Abdominal wall defect (specify:_____)
☐ Abnormal abdominal situs
☐ Anorectal anomaly
☐ Bowel obstruction (specify:_____)
☐ Echogenic bowel
☐ Hepatomegaly
☐ Small stomach
☐ Spleen anomaly
☐ Other: _____

Brain and Skull
☐ Abnormal corpus callosum
☐ Abnormal skull shape (specify:_____)
☐ Aqueductal stenosis
☐ Brainstem anomaly
☐ Cerebellar anomaly (specify:_____)
☐ Craniosynostosis
☐ Cyst(s) (specify:_____)
☐ Encephalocele
☐ Holoprosencephaly
☐ Macrocephaly
☐ Megalencephaly
☐ Microcephaly
☐ Neuronal migration anomaly (specify:_____)
☐ Posterior fossa malformation (specify:_____)
☐ Ventriculomegaly/hydrocephalus
☐ Other: _____

Cardiovascular
☐ Arrythmia/conduction defect
☐ Cardiomyopathy
☐ Congenital heart defect (specify:_____)
☐ Heterotaxy
☐ Vascular anomaly (specify:_____)
☐ Other: _____

Craniofacial
☐ Ear anomaly (specify:_____)
☐ Eye anomaly (specify:_____)
☐ Cleft lip
☐ Cleft palate
☐ Frontal bossing
☐ Micrognathia/retrognathia
☐ Midface hypoplasia
☐ Nose anomaly (specify:_____)
☐ Other: _____

Genital, Urinary and Renal
☐ Bladder anomaly (specify:_____)
☐ Genital anomaly (specify:_____)
☐ Kidney anomaly (specify:_____)
☐ Other: _____

Lymphatic Effusion
☐ Ascites
☐ Hydrops
☐ Lymphangioma
☐ Pericardial effusion
☐ Pulmonary effusion
☐ Skin edema
☐ Other: _____

Clinical Information for Prenatal Exome Sequencing continued on page 4

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Patient Name: _____ Date of Birth: _____

CLINICAL INFORMATION (continued)

Musculoskeletal

- ☐ Abnormal mineralization
- ☐ Arthrogryposis
- ☐ Fracture
- ☐ Shortened long bones
- ☐ Arm anomaly (specify:_____)
- ☐ Hand anomaly (specify:_____)
- ☐ Leg anomaly (specify:_____)
- ☐ Foot anomaly (specify:_____)
- ☐ Other: _____

Neck, Chest, and Lungs

- ☐ Bell-shaped chest
- ☐ Congenital diaphragmatic hernia
- ☐ Pulmonary hypoplasia
- ☐ Short ribs
- ☐ TEF/esophageal atresia
- ☐ Other: _____

Vertebra/Spine

- ☐ Kyphosis
- ☐ Sacral agenesis
- ☐ Scoliosis
- ☐ Spina bifida
- ☐ Other: _____

Other

- ☐ Abnormal placenta
- ☐ Fetal anemia
- ☐ Intrauterine growth restriction
- ☐ Large for gestational age
- ☐ Oligo/anhydramnios
- ☐ Polyhydramnios
- ☐ Other: _____

Additional Clinical Information: _____

Family History/Pedigree: _____

ADDITIONAL GENE PANEL INFORMATION

Cleft and Craniofacial Gene Panel (288 genes):
ABCC9, ACSS2, ACTB, ACTG1, ADAMTSL4, AHDC1, ALPL, ALX1, ALX3, ALX4, AMELX, AMER1, AMMECR1, AMOTL1, ANKH, ANKRD11, ARHGAP29, ARSB, ASPH, ASXL1, ASXL3, B3GAT3, B3GLCT, BCOR, BMP2, BMP4, BMPR1B, BPNT2, BRAF, BRD4, C2CD3, CBFB, CCNQ, CD96, CDC45, CDH1, CDKN1C, CDON, CENPF, CEP164, CHD5, CHD7, CILK1, CNOT1, COG1, COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3, COLEC10, COLEC11, CPLANE1, CREBBP, CTNND1, CTSK, CYP26B1, DDX59, DHCR7, DHODH, DISP1, DLL1, DLX4, DPF2, DPH1, DVL1, DVL3, EDN1, EDNRA, EFNA4, EFNB1, EFTUD2, EHMT1, EIF4A3, EP300, ERF, ESCO2, ESRP2, EVC, EVC2, EYA1, FAM20C, FBN1, FGD1, FGF10, FGF8, FGF9, FGFR1, FGFR2, FGFR3, FLNA, FLNB, FOXE1, FOXI3, FRAS1, FREM1, FST, FTO, FZD2, GAS1, GDF11, GJA1, GLI2, GLI3, GNAI3, GNAS, GNPTAB, GPC3, GPC4, GRHL3, GSC, GTF2E2, GZF1, HDAC8, HIST1H1E, HNRNPK, HUWE1, HYAL2, HYLS1, IDS, IDUA, IFT122, IFT140, IFT43, IGF1R, IGF2, IHH, IL11RA, INPPL1, IRF6, IRX5, ISM1, JAG1, KAT6A, KAT6B, KDM1A, KDM6A, KIAA0586, KIF7, KMT2D, KRAS, LOXL3, LRP2, LTBP1, MAFB, MAP3K7, MASP1, MED13L, MED25, MEGF8, MEIS2, MID1, MKS1, MN1, MSX1, MSX2, MTX2, MYCN, MYMK, MYT1, NBAS, NECTIN1, NEDD4L, NIPBL, OFD1, P4HB, PAX1, PAX3, PAX7, PDE4D, PGM1, PHEX, PHF21A, PHF8, PIEZO2, PIGN, PJA1, PLCB4, PLCH1, PLEKHA5, PLEKHA7, PLOD3, POLR1A, POLR1B, POLR1C, POLR1D, POR, PORCN, PPP1R12A, PRRX1, PSAT1, PTCH1, PTDSS1, PTPN11, RAB23, RAD21, RAX, RBM10, RECQL4, RIPK4, ROR2, RPGRIP1L, RPL5, RSPRY1, RUNX2, RYK, SATB2, SCARF2, SCLT1, SCN4A, SEC24D, SEMA3E, SF3B2, SF3B4, SHH, SHOC2, SHROOM3, SIN3A, SIX1, SIX2, SIX3, SIX5, SKI, SLC25A24, SMAD2, SMAD3, SMAD4, SMAD6, SMARCA4, SMARCB1, SMC1A, SMC3, SMG9, SMO, SMS, SMURF1, SNRPB, SON, SOST, SOX11, SOX6, SOX9, SPECC1L, SPRY1, SPRY4, STAG2, STIL, SUFU, SUMO1, TBC1D32, TBX1, TBX22, TCF12, TCOF1, TFAP2A, TFAP2B, TGDS, TGFB1, TGFB2, TGFB3, TGFBR1, TGFBR2, TGIF1, TLK2, TMCO1, TOPORS, TP63, TRAF7, TRRAP, TWIST1, TWIST2, TXNL4A, UBE3B, USP9X, VAX1, VCAN, WASHC5, WDR19, WDR35, WNT5A, YAP1, YWHAE, ZEB2, ZIC1, ZIC2, ZNF462, ZSWIM6

CARDIOLOGY GENE PANELS:

Congenital Heart Diseases Panel (187 genes):
ABL1, ACTA2, ACTB, ACTC1, ACTG1, ACVR1, ACVR2B, ACVRL1, ADAMTS10, AK7, ALMS1, ANKS6, ARHGAP31, ARMC4, ATRX, B3GAT3, BBS1, BBS10, BBS2, BCL9L, BCOR, BMPR2, BRAF, C21ORF59, CACNA1C, CBL, CCDC103, CCDC11, CCDC114, CCDC151, CCDC39, CCDC40, CCDC65, CCNO, CDK13, CENPF, CFAP300, CHD4, CHD7, CITED2, COL2A1, CREBBP, CRELD1, CYR61, DHCR7, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAH1, DNAH11, DNAH5, DNAH8, DNAH9, DNAI1, DNAI2, DNAJB13, DNAL1, DRC1, DSG2, DSP, DTNA, EFTUD2, EIF2AK4, ELN, ENG, EVC[†] EVC2, FBN1, FBN2, FGFR2, FLNA, FLNB, FOXC1, FOXC2, FOXF1, FOXH1, G6PC3, GAS2L2, GAS8, GATA4, GATA5, GATA6, GDF1, GJA1, GJA5, GLI3, GPC3, HAND1, HES7, HRAS, HYDIN, INVS, JAG1, KCNJ2, KIF7, KRAS, LEFTY2, LMNA, LRRC56, LRRC6, MAP2K1, MAP2K2, MCIDAS, MED13L, MEGF8, MEIS2, MID1, MKKS, MKS1, MMP21, MRE11, MYCN, MYH6, NAT10, NEK8, NF1, NIPBL, NKX2-5, NKX2-6, NME8, NODAL, NOTCH1, NOTCH2, NPHP3, NR2F2, NRAS, NSD1, NTRK3, OFD1, PIH1D3, PIK3R2, PITX2, PKD1L1, PKD2, PPP1CB, PQBP1, PRKD1, PRKG1, PRRX1, PTPN11, RAF1, RAI1, RBM10, RIT1, RSPH1, RSPH3, RSPH4A, RSPH9, SALL4, SCN1B, SCN5A, SEMA3E, SHOC2, SHROOM3, SKI, SMAD2, SMAD6, SOS1, SOS2, SOX2, SOX7, SPAG1, SPEG, TAB2, TBX1, TBX20, TBX3, TBX5, TCAP, TCTN2, TFAP2B, TGDS, TGFB2, TGFBR2, TLL1, TTC25, TWIST1, UBR1, VCL, WDR35, ZFPM2, ZIC3, ZMPSTE24, ZMYND10, ZNF469

Heterotaxy Panel (114 genes):
ACTC1, ACVR2B, AK7, ALMS1, ANKS6, ARMC4, BBS1, BBS10, BBS2, BCL9L, BCOR, BRAF, C21ORF59, CBL, CCDC103, CCDC11, CCDC114, CCDC151, CCDC39, CCDC40, CCDC65, CCNO, CENPF, CFAP300, CHD7, CRELD1, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAH1, DNAH11, DNAH5, DNAH8, DNAH9, DNAI1, DNAI2, DNAJB13, DNAL1, DRC1, ELN, EVC, EVC2, FOXF1, FOXH1, GAS2L2, GAS8, GATA4, GATA6, GDF1, GJA1, GPC3, HES7, HRAS, HYDIN, INVS, JAG1, KIF7, KRAS, LEFTY2, LMNA, LRRC56, LRRC6, MAP2K1, MAP2K2, MCIDAS, MED13L, MEGF8, MEIS2, MKS1, MMP21, MRE11, NAT10, NEK8, NF1, NKX2-5, NKX2-6, NME8, NODAL, NOTCH1, NOTCH2, NPHP3, NR2F2, NRAS, NSD1, OFD1, PIH1D3, PKD1L1, PKD2, PQBP1, PRRX1, PTPN11, RAF1, RIT1, RSPH1, RSPH3, RSPH4A, RSPH9, SCN5A, SHOC2, SHROOM3, SMAD2, SOS1, SPAG1, TBX1, TBX5, TCTN2, TTC25, UBR1, WDR35, ZIC3, ZMPSTE24, ZMYND10

RASopathy/Noonan Spectrum Disorder Panel (31 genes):
A2ML1, ACTB, ACTG1, BRAF, CBL, CDC42, HRAS, KAT6B, KRAS, LZTR1, MAP2K1, MAP2K2, NF1, NF2, NRAS, NSUN2, PPP1CB, PTEN, PTPN11, RAF1, RASA1, RASA2, RIT1, RRAS, SHOC2, SOS1, SOS2, SPRED1, TBCK, TSC1, TSC2

[†]For the Cardiology Gene Panels, Del/Dup analysis of the following genes is not available at this time: *A2ML1, ABL1, ACTA2, ACTG1, ACVR1, ACVR2B, ARHGAP31, BCL9L, CACNA1C, CCDC114, CDC42, CDK13, CHD4, CYR61, DSG2, DTNA, FOXH1, GATA5, GDF1, HAND1, LEFTY2, LZTR1, MAP2K1, MCIDAS, MID1, MRE11, MYH6, NAT10, NTRK3, PPP1CB, PRKD1, PRKG1, PRRX1, RASA2, RIT1, RRAS, SCN1B, SHROOM3, SKI, SMAD2, SMAD6, SOS2, TCAP, SPEG1, TFAP2B, TLL1, VCL, WDR35,* and *ZMYND10.*

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Patient Name: _____ **Date of Birth:** _____

SPECIMEN REQUIREMENTS

Tissue Testing:

20-30 mg in media or on a piece of sterile saline gauze (specimen should not be floating in saline).

*Please note: When requested, original POC tissue can be returned after testing is completed (if available). Please contact the lab at 513-636-4474 for details.

Prenatal Testing:

Amniotic Fluid: At least **25 mL** amniotic fluid is requested. Smaller samples are always accepted but may require additional culture time to meet minimum sample requirements. If multiple tests are being ordered, sending additional amniotic fluid may avoid delays related to a need to culture cells.

*Please note:

- In order to perform SNP Microarray testing on direct amniotic fluid samples (without culturing the cells), we require 25 mL of amniotic fluid. If the sample is sufficient, we will automatically perform SNP Microarray on direct amniotic fluid samples. However, bloody samples (fluid or cell pellet), low volume/low cell count samples, and/or samples with additional special study orders may need to be cultured to obtain SNP Microarray results.
- Amniotic fluid chromosome or microarray order includes (with additional charges): AF-AFP if gestational age 13W0D—36W6D with reflex to ACHE if AFP is abnormal. Order for ACHE will be added for the following indications: suspected or known neural tube defect, screen positive for neural tube defect, any open fetal lesions. AFP and ACHE will not be ordered for the following indications: fetal demise, twin reversed arterial perfusion (TRAP), twin-twin transfusion syndrome (TTTS), or any specimen type other than amniotic fluid.

CVS: 40 mg in sterile media. Smaller samples always accepted but may require additional culture time. **NO** formalin or freezing.

Parental samples:

- Prenatal Microarray: 5 mL blood in EDTA and 5 mL blood in NaHep OR one saliva kit for each parent.
- Exome sequencing: 5 mL blood in EDTA OR one saliva kit for each parent.
- ***Targeted variant testing by Sanger sequencing or aCGH: **Positive control samples are required for each variant.** If both parents are carriers for the same variant, positive controls from each parent are still required. 5 mL blood in EDTA OR one saliva kit for each positive control.
- Please note: We require confirmation of parental carrier status before testing the prenatal sample. If this is not possible, please call the laboratory to discuss acceptable alternatives.

****Backup cultures:**

- Prenatal Microarray orders: A backup culture will be held on an incubator for 5 business days after testing is complete. A backup culture will be frozen and stored for at least 1 year (2 years for patients with abnormal microarray results)
- Special study orders: A backup culture will be held on an incubator for 5 business days after testing is complete. A backup culture will be frozen and stored for at least 1 year.
- Chromosome orders: A backup culture will be held on an incubator for 7 days after testing is complete.

For questions about genetic testing specimen requirements, please call (513) 636-4474

Infectious Disease Testing:

At least 1 mL amniotic or body fluid in a sterile container, 1 mL of fetal blood in lavender top EDTA tube, or 0.3g fresh tissue in a sterile container.

For questions about infectious disease specimen requirements, please call (513) 636-9820

SHIPPING INFORMATION

Local courier is available; please call 513-636-4474 for information.

Shipping for samples that arrive **Monday-Friday:**

Cincinnati Children's
Genetics and Genomics Diagnostic Laboratory
3333 Burnet Ave.
TCHRF 1042
Cincinnati, OH 45229-3039

Shipping for samples that arrive on **Saturday** (Please call laboratory to inform):

Cincinnati Children's
Genetics and Genomics Diagnostics Laboratory
3333 Burnet Ave.
TCHRF 1042
DOCK 5
Cincinnati, OH 45229-3039

Be sure to mark the **Saturday** check box on the airbill