Cincinnati Children's
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GENETICS AND GENOMICS DIAGNOSTIC LABORATORY
For local courier service and/or inquiries, please contact 513-636-4474 • Fax: 513-636-4373
www.cincinnatichildrens.org/diagnosticlabs • Email: labgeneticcounselors@cchmc.org

Shipping Address:
3333 Burnet Avenue, Room R1042
Cincinnati, OH 45229-3039
Deliveries accepted Monday-Saturday

PEDIATRIC/ADULT GENETIC TEST REQUISITION

All Information Must Be Completed Before Sample Can Be Processed

PATIENT INFORMATION

Patient Name: _____, _____, _____
Last First MI

Address: _____

_____

Home Phone: _____

MR# _____ Date of Birth _____ / _____ / _____

Sex Assigned at Birth: ☐ Male ☐ Female ☐ Uncertain/Other:_____

SPECIMEN INFORMATION

SPECIMEN TYPE: ☐ Peripheral blood ☐ Skin biopsy (site): _____

☐ Bone Marrow ☐ Saliva ☐ Cytobrushes ☐ Other: _____

*See Page 3 for a list of acceptable specimen types for each test

Specimen Date: _____ / _____ / _____ Time: _____

Specimen Amount: _____

DRAWN BY: _____

INDICATIONS/DIAGNOSIS/ICD-10 CODE

☐ ADD/ADHD
☐ Acute myelogenous leukemia (AML)
☐ Amenorrhea: 1' or 2'
☐ Aplastic Anemia
☐ Autism Spectrum Disorder
☐ Broad thumbs and/or halluces
☐ Congenital heart anomaly
☐ Developmental Delay
☐ Dysmorphic features
☐ Encephalopathy
☐ Eye anomaly
☐ Erythematous "butterfly" lesion on face
☐ Failure to thrive
☐ Hydrocephalus
☐ Hyper/Hypopigmentation
☐ Hypotonia
☐ Immune deficiency
☐ Intellectual disability
☐ Language disorder
☐ Limb malformation
☐ MRI, abnormal
☐ Macrocephaly
☐ Microcephaly
☐ Myelodysplastic syndrome (MDS)
☐ PDD-NOS
☐ Pancytopenia
☐ Seizures, convulsions
☐ Short stature
☐ Other: _____

Newborn Indications:
☐ Abnormal NIPT/prenatal screen
☐ Suspected trisomy 21
☐ Suspected Turner's syndrome
☐ Ambiguous genitalia
☐ Other: _____

Family History
☐ Family history of genetic condition:
_____
_____
☐ Consanguinity (describe relationship):
_____
_____
☐ Known Chromosome Abnormality:
_____

BILLING INFORMATION (Choose ONE payment method)

☐ REFERRING INSTITUTION

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

☐ COMMERCIAL INSURANCE*

Insurance can only be billed if requested at the time of service.

Policy Holder Name: _____
Gender: _____ Date of Birth _____ / _____ / _____
Authorization Number: _____
Insurance ID Number: _____
Insurance Name: _____
Insurance Address: _____
City/State/Zip: _____
Insurance Phone Number: _____

*** 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.

PROVIDER INFORMATION

Provider Name (print): _____
Address: _____
Phone: (_____) _____ Fax: (_____) _____
Email: _____
Genetic Counselor/Lab Contact Name: _____
Phone: (_____) _____ Fax: (_____) _____
Email: _____

_____ Date: ____/ _____/____
Referring Physician Signature (REQUIRED)

Contact information for results/questions (if different than ordering provider):
Name and Title: _____
Phone: (_____) _____ Fax: (_____) _____
Email: _____

ETHNIC/RACIAL BACKGROUND (Choose All)

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

☐ **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.

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REQUIRED: Patient Name: _____ Date of Birth: _____

TEST(S) REQUESTED

Cytogenetic Testing

Chromosome Analysis

☐ Routine chromosome analysis*
- ☐ Chromosome mosaicism study*
- ☐ Reflex to SNP Microarray if chromosome results are normal†

☐ High resolution chromosome analysis*
- ☐ Chromosome mosaicism study*
- ☐ Reflex to SNP Microarray if chromosome results are normal†

*For chromosome analysis: reflex STAT prelim results on infants <1 month. Reflex to mosaicism study when sex chromosome/mosaic aneuploidy abnormality suspected by laboratory based on indications provided.
†Additional charge for reflex testing. If SNP Microarray is denied by insurance, Chromosome Analysis will be performed as the first test in the algorithm.

Microarray

☐ SNP Microarray - Constitutional
☐ Episignature Complete Analysis
☐ Episignature Targeted Analysis: Specify episignature†: _____

†*Please see Episignature Analysis test information sheet for available conditions*

Optical Genome Mapping

☐ Optical Genome Mapping (Genome-wide)
☐ Optical Genome Mapping† - Targeted Analysis : Known SV, gene and/or specific region: _____

† *Please contact GGDL to confirm OGM's coverage for the target region before ordering*

FISH (Fluorescent In Situ Hybridization)

☐ 22q11.2 del (VCFS) (metaphase FISH)
☐ SRY (Xp11.1q11.1/Yp11.2) (metaphase FISH)
☐ X/Y centromeres (Xp11.1q1.1/Yp11.1q11.1) (interphase FISH)
☐ Other FISH (please call lab): _____

Other Testing

☐ Special study: _____
☐ Cell Culture, storage & freezing
☐ Other: _____

Cytogenetic and Molecular Genetic Testing

Neurodevelopmental Reflex Genetic Test**

Tests will be run sequentially based on your selection below:

☐ Patient is macrocephalic: SNP Microarray → Fragile X → *PTEN*
☐ Male patient with normal or small head circumference: SNP Microarray → Fragile X
☐ Female patient with normal or small head circumference: SNP Microarray → Fragile X → *MECP2*

**If SNP Microarray is denied by insurance, Chromosome Analysis will be performed as the first test in the algorithm. See page 3 for additional information.

Chromosome Breakage Disorders Testing

☐ Bloom Syndrome - Sister Chromatid Exchange (SCE) analysis
☐ Chromosome Breakage Disorders Gene Sequencing Panel
(ATM, BLM, BRCA1, BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, LIG4, MAD2L2, MYSM1, NBN, NHEJ1, NSMCE3, PALB2, RAD51, RAD51C, RFWD3, SLX4, UBE2T, XRCC2)

Fanconi Anemia Testing

☐ Fanconi Anemia (FA) Chromosome Breakage Study
☐ Fanconi Anemia Gene Sequencing Panel
(BRCA1, BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, MAD2L2, PALB2, RAD51, RAD51C, RFWD3, SLX4, UBE2T, XRCC2)

If **both** FA Breakage Study and FA Gene Seq Panel are ordered, testing will be run sequentially *(breakage study then molecular sequencing if breakage study is positive; if breakage study is negative, molecular sequencing will not be performed)* unless concurrent testing is selected here:
☐ **Concurrent FA testing is requested**

Single Gene Sequencing

☐ *FANCA* full gene sequencing ☐ *FANCG* full gene sequencing
☐ *FANCC* full gene sequencing

Molecular Genetic Testing

☐ *ABCD1* gene sequencing (X-Linked Adrenoleukodystrophy)
- ☐ Reflex to *ABCD1* deletion/duplication by MLPA

☐ *ABCD1* deletion/duplication by MLPA
☐ 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
- ☐ **REFLEX to Whole Exome Sequencing††** (See additional details below)

☐ DNA Extraction and Storage
☐ Fragile X DNA testing
☐ *MECP2* sequence analysis (Rett syndrome)
☐ *MECP2* deletion/duplication analysis by MLPA
☐ Prader-Willi/Angelman - by methylation-sensitive MLPA
☐ *PTEN* Autism Spectrum Disorder sequencing
☐ Rubinstein-Taybi and Related Syndromes Gene Panel
(CREBBP, EP300, HNRNPH1, HNRNPH2, SIN3A, SIN3B, SRCAP with CREBBP and EP300 deletion/duplication analysis by MLPA)
- ☐ **REFLEXto Whole Exome Sequencing††** (See additional details below)

☐ Spinal Muscular Atrophy - *SMN1/SMN2* Copy Number Analysis by MLPA

††Whole exome sequencing (WES) orders require 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.

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REQUIRED: Patient Name: _____ Date of Birth: _____

TEST(S) REQUESTED CONTINUED

☐ Stickler Syndrome Gene Panel (13 genes)
BMP4, COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3, GZF1, LOXL3, LRP2, PLOD3, SOX9, VCAN
☐ Reflex to Cleft and Craniofacial Gene Panel
☐ **REFLEX to Whole Exome Sequencing††** (See additional details below)

☐ Treacher Collins Syndrome and Mandibulofacial Dysostosis Gene Panel (10 genes) *DHODH, EDNRA, EFTUD2, POLR1A, POLR1B, POLR1C, POLR1D, SF3B4, TCOF1, TXNL4A*
☐ Reflex to Cleft and Craniofacial Gene Panel
☐ **REFLEX to Whole Exome Sequencing††** (See additional details below)

☐ Other: _____

††Whole exome sequencing (WES) orders require 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.

CUSTOM GENE SEQUENCING

Gene(s) to be analyzed (specify): _____

_____

Only genes with clear published functional relationship to rare diseases are accepted.

Suspected syndrome/ condition: _____

Please choose one of the following:

☐ Full gene(s) sequencing
☐ Full gene(s) sequencing with reflex to deletion and duplication analysis, if indicated **(please see list of genes available for del/dup at www.cincinnatichildrens.org/deldup)**
☐ Familial mutation analysis
Proband's name: _____
Proband's DOB: _____
Proband's mutation: _____
Patient's relation to proband: _____
If testing was **not** performed at CCHMC, please include proband's report and at least 100ng of proband's DNA to use as a positive control.

DELETION AND DUPLICATION ASSAY

Gene(s) to be analyzed (specify): _____

_____

Please see list of available genes at: www.cincinnatichildrens.org/deldup

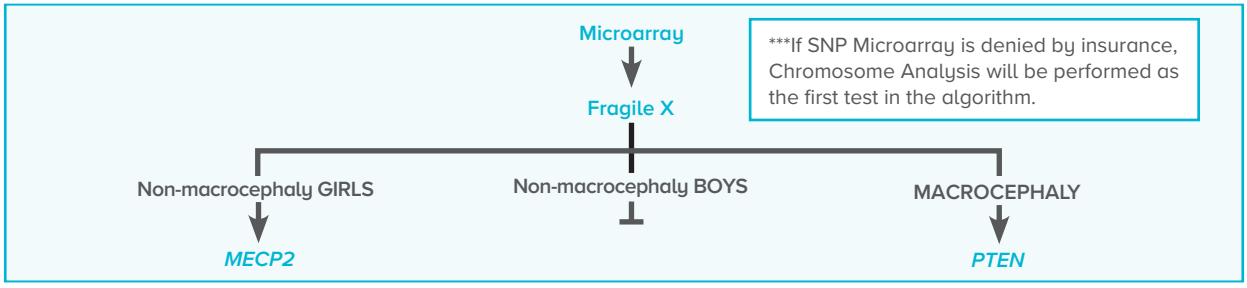
Suspected syndrome/ condition: _____

Please choose one of the following:

☐ Deletion and duplication analysis of gene(s) specified above
☐ Deletion and duplication analysis of gene(s) specified above with reflex to sequencing, if indicated
☐ Analysis of gene(s) specified above from previously analyzed deletion and duplication
☐ Familial deletion analysis
Proband's name: _____
Proband's DOB: _____
Proband's mutation: _____
Patient's relation to proband: _____
If testing was **not** performed at CCHMC, please include proband's report and at least 100ng of proband's DNA to use as a positive control.

NEURODEVELOPMENTAL REFLEX GENETIC TESTING ALGORITHM

Tests will be performed sequentially based on the path that your patient follows in the Neurodevelopmental Reflex Test algorithm. Charges will apply to the tests completed in the patient's defined Neurodevelopmental Reflex Test algorithm. Testing will only proceed to the next step if the previous test result is uninformative.



SPECIMEN REQUIREMENTS

- **Routine & High Resolution Chromosome Analysis:**
  3–5 mL blood (NaHep)
- **Optical Genome Mapping (OGM) Genome-wide and Targeted Analysis:**
  3 mL blood (NaHep) or (EDTA), tissue (1cm x 1cm), or punch biopsy (2mm tissue in sterile transport media or saline)
- **SNP Microarray:**
  3 mL blood (NaHep) and 3 mL blood (EDTA)
- **FISH Tests:** 1–3 mL blood (NaHep)
- **Neurodevelopmental Reflex Genetic Testing:**
  3 mL blood (NaHep) **and** 3–5 mL blood (EDTA)
- **Fragile X DNA Testing:**
  3 mL blood (EDTA)
- **Fanconi Anemia Chromosome Breakage Study:**
  5–10 mL blood (NaHep), 5–10 mL bone marrow (NaHep), or skin biopsy (3-4 mm tissue in sterile transport media)
- *ABCD1* del/dup by MLPA, EpiSignature Complete, EpiSignature Targeted, *MECP2* del/dup by MLPA, Prader-Willi/Angelman by MLPA, Spinal Muscular Atrophy - *SMN1/SMN2* Copy Number Analysis & Deletion/Duplication Assay:
  3 mL blood (EDTA)
- ***ABCD1, FANCA, FANCC, FANCG, MECP2, PTEN* & Custom Gene Sequencing:**
  3 mL blood (EDTA), saliva collection kit*, or 6 cytobrushes
- **Bloom syndrome – Sister Chromatid Exchange (SCE) analysis:**
  3–5 mL blood (NaHep)
- **Cleft and Craniofacial, Chromosome Breakage Disorders, Fanconi Anemia, Rubinstein-Taybi and Related Syndromes, Stickler Syndrome & Treacher Collins Syndrome and Mandibulofacial Dysostosis Gene Panels:**
  3 mL blood (EDTA) or saliva collection kit*

**Call the office at 513-636-4474 to obtain saliva collection kits*