

### 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		SPECIMEN INFORMATION		
Patient Name:,,,			od Skin biopsy (site): bbrushes Other: specimen types for each test	
Home Phone:  MR# Date of Birtt  Sex Assigned at Birth:		Specimen Amount:	/ Time:	
	INDICATIONS/DIA	GNOSIS/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	e ONE payment method)		ER INFORMATION	
Institution:  Address:  City/State/Zip:  Accounts Payable Contact Name:  Phone:  Fax:  Email:  COMMERCIAL INSURANCE* Insurance can only be billed if requester  Policy Holder Name:  Gender:  Date of Birth  Authorization Number:	ed at the time of service.	Address: Phone: ( ) Email: Genetic Counselor/Lab Contact I Phone: ( ) Email:  Referring Physician Signature (RE	estions (if different than ordering provider):  Fax: ( )	
Insurance ID Number: Insurance Name: Insurance Address: City/State/Zip: Insurance Phone Number:		☐ European American (White) ☐ Native American or Alaskan ☐ Pacific Islander	ACKGROUND (Choose All)  African-American (Black)  Asian-American  Ashkenazi Jewish ancestry	
<ul> <li>PLEASE NOTE:</li> <li>We will not bill Medicaid, Medicaid HMO, of CCHMC Patients, CCHMC Providers, or De</li> <li>If you have questions, please call 1-866-45</li> </ul>	esignated Regional Counties.	☐ Latino-Hispanic		

#### ☐ 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.



REQUIRED: Patient Name:	Date of Birth:
REGUINED. I ducit Haine.	2 440 0. 2.1411

GROAL BROAL SANCE, FANCE, F	TEST(S) REG	EQUESTED		
Chromosome Analysis	Cytogenetic Testing	Fanconi Anemia Testing		
□Record Anemia Gene Sequencing Panel				
GRIDAL SEAL BRICE_DECC, FANCE, FAN	☐ Routine chromosome analysis*			
S.X.A. UBEJT, XSCC.D)  □ fight resolution chromosome analysis* □ Chromosome mosoicism study* □ Reflex to SOM Microarray if chromosome results are normal*  For chromosome anelysis reflex.STAT prelim results on infents -1 month. Reflex to mosacless developmentally suspected by absorbation years and electrons provided as the first test in the algorithm.  ■ SNP Microarray is represented as the first test in the algorithm.  ■ SNP Microarray is performed as the first test in the algorithm.  ■ Chromosome Analysis will be performed as the first test in the algorithm.  ■ Pipasature Complete Analysis is peorly episignature*.  □ Pipasature Confeder Analysis is peorly episignature	☐ Chromosome mosaicism study*	. •		
High resolution chromosome analysis:	☐ Reflex to SNP Microarray if chromosome results are normal <sup>†</sup>			
Reflex to SNP Microarray if chromosome results are normal?   Fire-rithmosome analysis: reflex. FTAT prelim results on infants of month. Reflex to mosalcions subjected by laboratory based on indications provided. Additional change for reflex testing. If SNP Microarray is denied by insurance. Chromosome Analysis will be performed as the first test in the algorithm.   Microarray	☐ High resolution chromosome analysis*			
The chromosome analysis reflex STAT prelim results on intents of month. Beflex 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, Comonsome Analysis will be performed as the first test in the algorithm.	•			
Single Gene Sequencing   FANCG full gene fall gene full gene sequencing   FANCG full gene fall gene	<u>,                                      </u>	<u>not</u> be performed) unless concurrent testing is selected here:		
Single-dends Special Complete Analysis in SPM 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 Complete Analysis   Episignature Complete Analysis   Episignature Targeted Analysis: Specify episignature*:   Reflect No. ABCD1 deletion/duplication by MLPA		Concurrent FA testing is requested		
General Section	suspected by laboratory based on indications provided.	· · ·		
Molecular Genetic Testing     SNP Microarray - Constitutional     Episignature Complete Analysis: Specify episignature!     Filease see Episignature Analysis test information sheet for available conditions     Filease see Episignature Analysis test information sheet for available conditions     Optical Genome Mapping     Optical Genome Paralle Mapping     Optical Genome Paralle Mapping     Opt				
SNP Microarray - Constitutional   Chispiagnature Complete Analysis   Reflex to ABCD1 deletion/duplication by MLPA   Reflex to ABCD2 deletion/duplication by MLPA   Reflex to ABCD1 deletion/duplication by MLPA   Reflex to ABCD1 deletion/duplication analysis b	Chromosome Analysis will be performed as the first test in the algorithm.	☐ FANCC full gene sequencing		
Episignature Complete Analysis     Episignature Targeted Analysis: Specify episignature <sup>1</sup> ;     Episignature Targeted Analysis: Specify episignature <sup>2</sup> ;     Episignature Targeted Analysis: Specify episignature <sup>3</sup> ;     Episignature Targeted Analysis: Specify episignature <sup>3</sup> ;     Episignature Targeted Analysis: Specify episignature <sup>3</sup> ;     Episignature Analysis test information sheet for evallable conditions     Cleft and Craniofacial Gene Panel (288 genes)     ABCD7 deletion/duplication by MLPA     ABCD7 deletion/duplication by MLPA     Cleft and Craniofacial Gene Panel (288 genes)     ABCS9 ACS\$2.ACT8.ACT67.AMMC.RAM.RAD1.AMMC.RAM.RAD1.AMMC.RAM.RAD1.AMMC.RAM.RAD1.AMMC.RAM.RAD1.AMMC.RAM.RAD2.CAMMC.RAM.RAD1.AMMC.RAM.RAD8.AMMC.RAM.RAM.RAD8.AMMC.RAM.	Microarray	Molecular Genetic Testing		
Episignature Targeted Analysis: Specify episignature <sup>1</sup> .		☐ ABCD1 gene sequencing (X-Linked Adrenoleukodystrophy)		
Pilease see Episignature Analysis test information sheet for available conditions		☐ Reflex to ABCD1 deletion/duplication by MLPA		
ABCCS, ACSS2, ACTB, ACTG1, ADAMTSL4, AHDC1, ALPL, ALX1, ALX3, ALX5 AMEX, AMERT, AMERT, AMEX, AM	Episignature Targeted Analysis: Specify episignature :	☐ ABCD1 deletion/duplication by MLPA		
Optical Genome Mapping  □ Optical Genome Mapping (Genome-wide) □ Optical Genome Mapping (Genome-wide) □ Optical Genome Mapping (Frangeted Analysis: Known SV, gene and/or optical Genome Mapping) - Targeted Analysis: Known SV, gene and/or specific region: □ Optical Genome Mapping (Frangeted Analysis: Known SV, gene and/or optical Genome Mapping) - Targeted Analysis: Known SV, gene and/or specific region: □ Please contact GGDL to confilm OGM's coverage for the target region before ordering optical Genetic Testing (France) □ 12 optical Genome Mapping) - Targeted Analysis: Known SV, gene and/or specific region: □ 12 optical Genome Mapping) - Targeted Analysis: Known SV, gene and/or specific region: □ 12 optical Genome Mapping) - Targeted Analysis: Known SV, gene and/or specific region: □ 12 optical Genome Mapping) - Targeted Analysis: Known SV, gene and/or specific region: □ 12 optical Genome Mapping) - Targeted Analysis: Known SV, gene and/or specific region: □ 12 optical Genome Mapping) - Targeted Analysis: Known SV, gene and/or specific region: □ 12 optical Genome Mapping) - Targeted Analysis: Known SV, gene and/or specific region: □ 12 optical Genome Mapping) - Targeted Analysis: Known SV, gene and/or specific region: □ 12 optical Genome Mapping) - Targeted Analysis: Known SV, gene and/or specific region: □ 22 optical Study: □ 22 optical Study: Specific Region Study: Specific Reg	†Please see Episignature Analysis test information sheet for available conditions	☐ Cleft and Craniofacial Gene Panel (288 genes)		
Optical Genome Mapping (Genome-wide) □ Optical Genome Mapping (Fragreted Analysis: Known SV, gene and/or specific region: specific region: yPlease contact GOD. to confirm OGM's coverage for the target region before ordering  FISH (Fluorescent in Situ Hybridization) □ 22qtil. 2 del (VCFS) (metaphase FISH) □ 22qtil. 2 del (VCFS) (metaphase FISH) □ SPRY (Xpft.lqtt.lYrpt1.2) (metaphase FISH) □ SPRY (Xpft.lqtt.lYrpt1.2) (metaphase FISH) □ Other FISH (please call lab): □ Testing □ Other FiSH (please call lab): □ Clulture, storage & freezing □ Clulture, storage & freezing □ Other: □ Clulture, storage & freezing □ Other: □ Patient is macrocephalic: SNP Microarray → Fragile X → PTEN □ Male patient with normal or small head circumference: SNP Microarray → Fragile X → MECP2 □ Microarray → Fragile X → PTEN □ Chromosome Breakage Disorders Testing □ Chromosome Breakage Disorders Gene Sequencing Panel □				
□ Optical Genome Mapping¹ - Targeted Analysis : Known SV, gene and/or specific region:       CDON, CENPF, CEP164, CHD5, CHD7, CULX1, CNOT1, CO31, CO1.1412, CO1.44, CO1.942, CO1.942, CO1.942, CO1.942, CO1.943, CO1.942, CO1.943, CO1.9		ASPH, ASXL1, ASXL3, B3GAT3, B3GLCT, BCOR, BMP2, BMP4, BMPR1B		
Specific region:  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)  □ SPY (Xp11.1q11.1/Yp11.2) (metaphase FISH)  □ SYY (Xp11.1q11.1/Yp11.2) (metaphase FISH)  □ Other FISH (please call lab): □ Other FISH (please call lab): □ Chira Special study: □ Cell Culture, storage & freezing  □ Chira Study: □ Cell Culture, storage & freezing  □ Other: □ Special study: □ Cell Culture, storage & freezing □ Special study: □ Cell culture, storage & freezing □ Other: □ Special study: □ Cell culture, storage & freezing □ Special study: □ S				
*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.Yp11.2) (metaphase FISH) □ SRY (Xp11.1q11.Yp11.2) (metaphase FISH) □ Other FISH (please call lab): □ Other FISH (please call lab): □ Other FISH (please call lab): □ Other Testing □ Cell Culture, storage & freezing □ Special study: □ Cell Culture, storage & freezing □ Yytogenetic and Molecular Genetic Testing  Neurodevelopmental Reflex Genetic Testing  Neu		COL11A2, COL2A1, COL9A1, COL9A2, COL9A3, COLEC10, COLEC11		
FISH (Fluorescent in Situ Hybridization)    SPACE   Page				
FISH (Fluorescent in Situ Hybridization)	r rouge contact copie to commit com a coverage for the target region points atteming			
□ 22q11.2 del (VCFS) (metaphase FISH) □ SRY (Xp11.4q1.1/Yp11.2) (metaphase FISH) □ SRY (Xp11.4q1.1/Yp11.2) (metaphase FISH) □ Other FISH (please call lab): □ Other FISH (please call lab): □ Other Testing Other Testing □ Special study: □ Special study: □ Cell Culture, storage & freezing □ Other: □ Starta A Fare & Fish, Picz Prot., Port., Port.	EICH (Fluorescent In City Lhybridization)			
□ SRY (Xp11.q11.1/Yp11.2) (metaphase FISH) □ XY centromeres (Xp11.q1.1/Yp11.1q1.1) (interphase FISH) □ XY centromeres (Xp11.q1.1/Yp11.1q1.1) (interphase FISH) □ Other FISH (please call lab): □ Other FISH (please call lab): □ Special study: □ Cell Culture, storage & freezing □ Cell Culture, storage & freezing □ Cell Culture, storage & freezing □ Cher: □ Special study: □ Cell Culture, storage & freezing □ Special study: □ Cell Culture, storage & freezing □ Special study: □ Cell Culture, storage & freezing □ Special study: □ Cell Culture, storage & freezing □ Cher: □ Sylva Microarray → Fragile X → MECP2 Neurodevelopmental Reflex Genetic Testing Neurodevelopmental Reflex Genetic Testing Neurodevelopmental Reflex Genetic Testing Nals patient with normal or small head circumference: Sylva Microarray → Fragile X → MECP2 □ Patient is macrocephalic: Sylv Microarray → Fragile X → PTEN □ All patient with normal or small head circumference: Sylva Microarray → Fragile X → MECP2 □ MECP2 sequence analysis (Rett syndrome) □ Chromosome Breakage Disorders Gene Sequencing Panel □ Chromosome Breakage Disorders Gene Sequencing Panel □ Chromosome Breakage Disorders Festing □ Rubinstein-Taybi and Related Syndrownes Gene Panel □ (CREBBP, Er300, HNRNPH), HNRNPH2, SINJA, SINJB, SRCAP with CREBBP and EP300 deletion/duplication analysis by MLPA)				
□ X/Y centromeres (Xp11.1q1.1/Yp11.1q11.1) (interphase FISH) □ Other FISH (please call lab): □ Other FISH (please call lab): □ Other Testing ○ Other Testing □ Special study: □ Special study: □ Cell Culture, storage & freezing □ Other: □ Cell Culture, storage & freezing □ Other: □ She				
MEISZ, MID1, MKS1, MN1, MSX1, MSX2, MTX2, MYCN, MYMK, MYT1, NBAS, NECTIN1, NEODAL, NIPBL, OFD1, P4HB, PAX1, PAX3, PAX7, PD4D, PGM PHEX, PHF31A, PHF31, PICTO, PO1, P4HB, PAX1, PAX3, PAX7, PD4D, PGM PHEX, PHF31A, PHF31A, PHF31A, PLCB4, PLCH1, PLEKHA5, PLCB1, PD41A, PD41B, PAX1, PAX3, PAX7, PD42B, PAX1, PAX3, PAX1, PD42B, PAX1, PAX3, PAX1, PB42B, PAX1, P				
Other Testing  □ Special study: □ Special study: □ Cell Culture, storage & freezing □ Cell Culture, storage & freezing □ Cell Culture, storage & freezing □ Chercian Molecular Genetic Testing  Neurodevelopmental Reflex Genetic Testing Neurodevelopmental Reflex Genetic Testing Neurodevelopmental Reflex Genetic Testing □ 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 → 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 Gene Sequencing Panel (ATM, BLM, BRCA1, BRCA2, BRIP, ERCC4, FANCA, FA	☐ Other FISH (please call lab):			
□ Special study: □ Cell Culture, storage & freezing □ Cherria	011 7 11	NECTIN1, NEDD4L, NIPBL, OFD1, P4HB, PAX1, PAX3, PAX7, PDE4D, PGM1		
□ Cell Culture, storage & freezing □ Other: □ O		PLEKHA7, PLOD3, POLR1A, POLR1B, POLR1C, POLR1D, POR, PORCN		
SATB2, SCARF2, SCLT1, SCN4A, SEC24D, SEM3E, SF3B2, SF3B2, SH3B, SHH SHOC2, SHROOM3, SIN3A, SIN1, SIX2, SIX3, SIX5, SKI, SKIX, SKIX6, SKIX, SMG9, SMO Sytogenetic 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 Gene Sequencing Panel (ATM, BLM, BRCA1, BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC, FANCD, FANCE, FANCE				
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, TGFB1, TGFBR2, TGIF1 TLL2, TMCO1, TOPORS, TP63, TRAF7, TRRAP, TWIST1, TWIST2, TXNL4A UBE3B, USP9X, VAX1, VCAN, WASHC5, WDR19, WDR35, WNT5A, YAP1 YWHAE, ZEB2, ZIC1, ZIC2, ZNF462, ZSWIM6 SNP Microarray → Fragile X Female patient with normal or small head circumference: SNP Microarray → Fragile X → MECP2  "If 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  Chromosome Breakage Disorders Gene Sequencing Panel (ATM, BLM, BRCA1, BRCA2, BRIP1, ERCC4, FANC6, FANC6, FANC6, FANC6, FANC7, FANC6, PANC7, PANC6, DEW/N3, NIM, NSMC63, MARCB1, SMC3, SMC9, SMC1, SMC3, SMC9, SM		SATB2, SCARF2, SCLT1, SCN4A, SEC24D, SEMA3E, SF3B2, SF3B4, SHH		
Sytogenetic and Molecular Genetic Testing  Neurodevelopmental Reflex Genetic Test**  Neurodevelopmental Reflex Genetic Test**  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  □ Permale patient with normal or small head circumference:  SNP Microarray → Fragile X → MECP2  **If 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  □ Chromosome Breakage Disorders Gene Sequencing Panel  (ATM, BLM, BRCA1, BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANC	Li Other			
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 □ Premale patient with normal or small head circumference: □ SNP Microarray → Fragile X □ Premale 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 □ Chromosome Breakage Disorders Gene Sequencing Panel (ATM, BLM, BRCA1, BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC2, FANCD2, FANCE, FANCB, FANCB, FANCB, FANCB, FANCB, FANCB, FANCB, WISM, NEM, NEM, NEMCE3, PANCB, PANCB, FANCB, FANCB, LIG4, MAD2L2, MYSMI, NBN, NHEJ1, NEMCE3, USPS, VAX1, VCAN, WASHCS, WDR19, WDR35, WDR19, WDR35, WDR19, WDR19, WDR19, WDR19, WDR19, WDR19, TXND4  TEST WDR19, WDR19, WDR35, WDR19, WDR19, WDR19, WDR11, TXND4B, USPS, TXND4  UBE38, USPS, VAX1, VCAN, WASHCS, WDR19, WDR35, WDR36, WDR36, WDR36, WDR36, WDR19, WDR35, WDR19, WDR36, WDR19, WDR36, WDR19, WDR36, WDR19, WDR35, WDR19, WDR35, WDR19, WDR35, WDR19, WDR35, WDR19, WDR35, WDR19, WDR35, WDR19, WDR36, WDR1	Cytogenetic and Molecular Genetic Testing	SMS, SMURF1, SNRPB, SON, SOST, SOX11, SOX6, SOX9, SPECC1L, SPRY1 SPRY4, STAG2, STIL, SUFU, SUMO1, TBC1D32, TBX1, TBX22, TCF12, TCOF1		
□ 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: □ Patient with normal or small head circumference: □ Patient with normal or small head circumference: □ Pragile X □ DNA Extraction and Storage □ Fragile X DNA testing □ MECP2 sequence analysis (Rett syndrome) □ MECP2 sequence analysis (Rett syndrome) □ MECP2 deletion/duplication analysis by MLPA □ Prader-Willi/Angelman - by methylation-sensitive MLPA □ Prader-Willi/Angelman - by methylation-sensitive MLPA □ Prader-Willi/Angelman Related Syndromes Gene Panel (ATM, BLM, BRCA2, BRIP1, ERCC4, FANCB, FANC	Neurodevelopmental Reflex Genetic Test** Tests will be run sequentially based on your selection below:	TLK2, TMCO1, TOPORS, TP63, TRAF7, TRRAP, TWIST1, TWIST2, TXNL4A		
□ Male patient with normal or small head circumference:       □ REFLEX to Whole Exome Sequencing <sup>#</sup> (See additional details below         SNP Microarray → Fragile X       □ DNA Extraction and Storage         □ Fragile X DNA testing       □ Fragile X DNA testing         SNP Microarray → Fragile X → MECP2       □ MECP2 sequence analysis (Rett syndrome)         **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.       □ MECP2 deletion/duplication analysis by MLPA         □ Prader-Willi/Angelman - by methylation-sensitive MLPA       □ PTEN Autism Spectrum Disorder sequencing         □ Chromosome Breakage Disorders Gene Sequencing Panel (ATM, BLM, BRCA1, BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCB, FANC				
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  □ Chromosome Breakage Disorders Gene Sequencing Panel  (ATM, BLM, BRCA1, BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE,		☐ REFLEX to Whole Exome Sequencing <sup>#</sup> (See additional details below		
□ 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  □ Chromosome Breakage Disorders Gene Sequencing Panel  (ATM, BLM, BRCA1, BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE,		□ DNA Extraction and Storage		
SNP Microarray → Fraqile 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.  **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.  **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.  **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.  **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.  **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.  **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.  **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.  **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.  **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.  **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.  **If SNP Microarray is deletion/duplication analysis by MLPA  **If SNP Microarray is	☐ Female patient with normal or small head circumference:	☐ Fragile X DNA testing		
performed as the first test in the algorithm. See page 3 for additional information.  □ Prader-Willi/Angelman - by methylation-sensitive MLPA  □ Prader-Willi/Angelman - by methylation-sensitive MLPA  □ PTEN Autism Spectrum Disorder sequencing  □ Chromosome Breakage Disorders Testing □ Chromosome Breakage Disorders Gene Sequencing Panel  (ATM, BLM, BRCA1, BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCF, FANCG, FANCI, LIG4, MAD2L2, MYSM1, NBN, NHEJ1,  NSMCF3, PALS2, PAD51, PAD51C, PEWD3, SLYA, LIBE37, YRCC2)	$\underline{\text{SNP Microarray}} \rightarrow \underline{\text{Fragile } X} \rightarrow \underline{\text{MECP2}}$			
□ Prader-Willi/Angelman - by methylation-sensitive MLPA  Chromosome Breakage Disorders Testing □ Chromosome Breakage Disorders Gene Sequencing Panel (ATM, BLM, BRCA1, BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCE, FANCE, FANCI, FANCI, LIG4, MAD212, MSM, SM, NHEJ1, NISMCER, PALB 2, PAD51, PAD51C, PEWD3, SLYA, LIBE2T, YPCC2)	**If SNP Microarray is denied by insurance, Chromosome Analysis will be	☐ MECP2 deletion/duplication analysis by MLPA		
□ Chromosome Breakage Disorders Gene Sequencing Panel  (ATM, BLM, BRCA1, BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, PANCB, BRANCI, BRANCI	periornieu as the first test in the algorithm. See page 3 for additional information.	☐ Prader-Willi/Angelman - by methylation-sensitive MLPA		
(ATM, BLM, BRCA1, BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC, FANCD2, (CREBBP, EP300, HNRNPH1, HNRNPH2, SIN3A, SIN3B, SRCAP with CREBBP and FANCE, FANCE, FANCE, FANCE, FANCE, FANCE, PAUR3, PAD51, PAD5	Chromosome Breakage Disorders Testing	☐ PTEN Autism Spectrum Disorder sequencing		
NSMCES PALES PADES PADES PERMOS SLYA LIBERT YPCCS)	(ATM, BLM, BRCA1, BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC, FANCD2,	(CREBBP, EP300, HNRNPH1, HNRNPH2, SIN3A, SIN3B, SRCAP with CREBBP and		

\*\*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 <a href="https://www.cincinnatichildrens.org/exome">www.cincinnatichildrens.org/exome</a> to obtain the required documents. WES testing will NOT be started until all forms are completed and received by the lab.

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



REQUIRED: Patient Name: Date of Birth:	REQUIRED: Patient Name:	Date of Birth:
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## **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)

$\square$ Treacher Collins Syndrome and Mandibulofacial Dysostosis Gene Par	ıel
(10 genes) DHODH, EDNRA, EFTUD2, POLR1A, POLR1B, POLR1C, POLR1D,	
SF3B4, TCOF1, TXNL4A	
Deflects Cleft and Cranicfacial Cons. Danal	

- □ Reflex to Cleft and Craniofacial Gene Panel
- ☐ REFLEX to Whole Exome Sequencing\* (See additional details below)

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"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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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
- $\square$  Familial deletion analysis

Proband's name: \_

Proband's DOB:

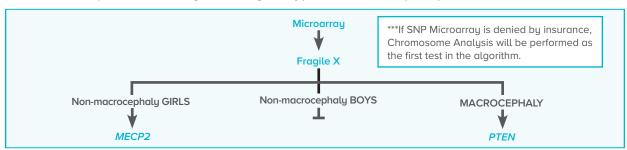
Proband's mutation: \_

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
- 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