

PHONE 1.800.411.4363 FAX 1.800.434.9850 CONNECT



# **SEQUENTIAL TRIO WHOLE EXOME SEQUENCING REQUISITION (TEST CODE: 1601)**

FASCIMILE INFORMATION				
			/	/
From			Date (MM	/ DD / YYYY) # Pages
PATIENT INFORMATION (COMPLETE OF	NE FORM FOR EACH PERSON TESTED)			
				/ /
Patient Last Name	Patient First Name		MI	Date of Birth (MM / DD / YYYY)
Address	City	State	Zip Genetic Sex:	Phone
Accession # Hos	pital / Medical Record #	Patient discharged from the hospital/facility:	Female	Male Unknown
		Yes No	Gender identity (if di	rrerent from above):
REPORTING RECIPIENTS				
Ordering Physician		Institution Name		
Email (Required for International Clients)		Phone	Fax	
ADDITIONAL RECIPIENTS				
Name		Email	Fax	
Name		Email	Fax	
PAYMENT (FILL OUT ONE OF THE OPTI	ONS BELOW)			
SELF PAYMENT				
<u> </u>	To Patient			
	io i diletti			
( INSTITUTIONAL BILLING				
Institution Name	Institution Code Inst	itution Contact Name Ins	stitution Phone	Institution Contact Email
INSURANCE				
<u> </u>	tient is Aware of Out-Of-Pocket Costs	(excludes prenatal testing)		
<del>_</del>		Diagnosis Code(s) 3. Name of Ordering	Physician / Inst	red Signature of Authorization
REQUIRED FIEND 1. copy of the	2. ICDTO	oraginosis code(s) 5. Name of ordering	1 Hysician 4. Ilisi	
No control of the control	///			///
Name of Insured	Insured Date of Birth (MM / DD / YYYY)	Name of Insured		Insured Date of Birth (MM / DD / YYYY)
Patient's Relationship to Insured	Phone of Insured	Patient's Relationship to I	nsured	Phone of Insured
		_ :		
Address of Insured		Address of Insured		
City	State Zip	— City		State Zip
	2.6			2.5
Primary Insurance Co. Name	Primary Insurance Co. Phone	Secondary Insurance Co.	Name	Secondary Insurance Co. Phone
Primary Member Policy #	Primary Member Group #	Secondary Member Policy	/#	Secondary Member Group #
By signing below, I hereby authorize B: claim. I understand that I am responsit by my insurance carrier for reasons in Genetics any and all payments that I rescreening tests.	ole for any co-pay, co-insurance, and c cluding, but not limited to, non-covere	unmet deductible that the insuranced and non-authorized services. I u	e policy dictates, inderstand that I	as well as any amounts not paid am responsible for sending Baylor
Patient's Printed Name	Patient's S	Signature		/ / /
	i diciti 3			2010 ( / 20 / 1111)



Maternal Sample (EDTA or Saliva) Paternal Sample (EDTA or Saliva)

Requisition

BAYLOR GENETICS 2450 HOLCOMBE BLVD. **SUITE 2210** HOUSTON, TX 77021-2024 PHONE 1.800.411.4363 FAX 1.800.434.9850

☐ Updated Clinical Notes/Summary

Pedigree

CONNECT



SEQUENTIAL TRIO WHOLE	EXOME SE	QUENCING	REQUISITION (T	EST CO	ODE: 1601)	
Patient Last Name	Patient First N	Name	MI		Date of Birth (MM / DD / YYYY)	Genetic Sex
TEST INFORMATION						
This testing is ONLY available as a follo number is indicated on the WES report version 1 or 2 Proband WES run previo details and requisition.	. If you are und	ertain which WE	S version number was	complete	ed, please call client services	to verify. If you had a
BIOLOGICAL PARENTS INFORMATION						
BIOLOGICAL PARENTS SAMPLES ARE F BEEN RECEIVED. PLEASE SHIP TOGETH		R SEQUENTIAL T	RIO WES TESTING. TES	TING CAN	INOT BE INITIATED UNTIL BO	TH PARENTAL SAMPLES HAVE
<ul> <li>If samples have been previously s is sufficient. If parental saliva san instructed below.</li> </ul>						
<ul> <li>If parental samples have NOT bee be faxed to 713-798-2787. Send 11 CHILD'S NAME.</li> </ul>						
MATERNAL INFORMATION						
Maternal Last Name		Maternal First N	ame		MI M	laternal Date of Birth (MM / DD / YYYY)
Sample previously submitted?	AMPLE TYPE:	NO, Please fi	ll out sample information	below.	YES, Baylor Genetic	s Lab #:
Asymptomatic		SAMPLE TYPE:	Blood		/ /	Not Available
Symptomatic (Attach summary of findi	ngs)		Saliva Buccal Swab	Date of	Collection (MM / DD / YYYY)	O To Be Sent Later *
PATERNAL INFORMATION						
Paternal Last Name		Paternal First Na	ame		MI P	aternal Date of Birth (MM / DD / YYYY)
Sample previously submitted?	AMPLE TYPE:	NO, Please fi	ll out sample information	below.	YES, Baylor Genetic	s Lab #:
Asymptomatic		SAMPLE TYPE:	Blood		/ /	Not Available
Symptomatic (Attach summary of findi	ngs)		Saliva Buccal Swab	Date of	Collection (MM / DD / YYYY)	To Be Sent Later *
ADDITIONAL REPORTING OPTIONS						
If a box is not checked the lab will defaul	t to No / Not R	eport.				
Option for Reporting of ACMG Secondar Variants in genes included in the ACMG s receive their own report on these finding	econdary find	ings guidelines v	will be reported for eac	h family r	nember marked below. Each	marked family member will
Proband Mother	Father					
Option for Reporting of Incidental Findir Pathogenic and likely pathogenic variant		ered under Cate	gory II of the Incidental	l Findinas	section of the consent form	will be reported.
Please report pathogenic and likely pa	-			_		
Trio and Quad Orders with Both Parents For variants in genes with no known dise	Only – Option	for Reporting o	of Research Findings		le novo.	
Please report biallelic and de novo var			·			
ITEM CHECKLIST						

Consent Form Signed By All Individuals Tested

Updated Indication for Study Checklist



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## **SEQUENTIAL TRIO WHOLE EXOME SEQUENCING REQUISITION (TEST CODE: 1601)**

Patient Last Na	me Patient First	Name		Date of Birth (MM / DD / Y	YYY) Genetic Sex
INDICATION F	OR TESTING (REQUIRED)				
Please provid	e the following clinical information reg with the corresponding HPO number (h ncing results. If the laboratory requires	ttp://human-phenotype	e-ontology.github.io/). Th	is information is needed to	o facilitate interpretation of whole
Physician Nam	e	Physician Phone		ICD-10 Diagnos	is Code(s)
PRE/PERINAT	FAL HISTORY	EYE DEFECTS & V	'ISION ······	MOTOR/COG  ☐ 0000750	NITIVE DEVELOPMENT  Delayed Speech & Language Development
0001511 0001562 0001561	Intrauterine Growth Restrictions Oligohydramnios Polyhydramnios	☐ 0000589 Col	ndness oboma ridia	<ul><li>☐ 0001270</li><li>☐ 0002376</li><li>☐ Intellectu</li></ul>	Delayed Motor Milestones  Developmental Regression al Disability
0000476 0000776 0001508	Cystic Hygroma Congenital Diaphragmatic Hernia Failure to Thrive	0000568 Mic			01256 Mild 02342 Moderate 0864 Severe
0001539 0002084 0010880	Omphalocele Encephalocele Increased Nuchal Translucency		abismus aract Congenital Bilateral	□ 0000729 □ □	Autistic Spectrum Disorder
STRUCTURAL	BRAIN ABNORMALITIES	NEUROLOGICAL		CRANIOFACIA	۱L
0001360	Holoprosencephaly	0001284 Are	flexia	0000256	Macrocephaly
0001339	Lissencephaly		leptic Encephalopathy		Microcephaly
0002084	Encephalocele Hydrocephalus		zures		Craniosynostosis Cleft Upper Lip
0000230	Ventriculomegaly	□ 0002373	Febrile Seizures	0000204	Cleft Palate
0001273	Abnormality of Corpus Callosum	□ 0012469	Infantile Spasms	0000316	Hypertelorism
0002539	Cortical Dysplasia	0002123	Generalized Myoclonic Seizures	0000601	Hypotelorism
0012444	Brain Atrophy		Generalized Tonic-clonic	0008050	Abnormality of the Palpebral Fissures
0002352	Leukoencephalopathy	0002069	Seizures	0000286	Epicanthal Folds
0002269	Abnormality of Neuronal Migration	0010818	Generalized Tonic Seizu	res 0000288	Abnormality of the Philtrum
0002126	Polymicrogyria	0010819	Atonic Seizures	0010938	Abnormality of the External Nose
0001302	Pachgyria	0002121	Absence Seizures		
0002500	Abnormality of Cerebral White Matter	0011169	Generalized Clonic Seizu	ıres	
0007266	Cerebral Dysmyelination	0001251	Ataxia		
0006808	Cerebral Hypomyelination	0001332	Dystonia		
0002134	Abnormality of the Basal Ganglia		Chorea		
0002363	Abnormality of the Brainstem	0001257	Spasticity		
0007360	Aplasia/Hypoplasia of the Cerebellum	0001237			
0006817	Aplasia/Hypoplasia of the Cerebellar Vermis		Neuropathy		

Indications continued on next page



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Date of the con-				/	/	Court C
Patient Last Na	me Patient First Na	ime	MI E	Date of Birth (MM	I / DD / YY	YY) Genetic Sex
INDICATION F	OR TESTING (REQUIRED) - CONTINUED					
HAIR & SKIN		CARDIAC		····· GEN	ITOURIN	4RY
0000957	Cafe-Au-Lait Spots	0001631	Atria Septal Defect		0000113	Polycystic Kidney Dysplasia
0001034	Hypermelanotic Macule	0001629	Ventricular Septal Defect		0000107	Renal Cyst
0001010	Hypopigmentation of the Skin	0001655	Patent Foramen Ovale		0008738	Partially Duplicated Kidney
0008066	Abnormal Blistering of the Skin			_	0000104	Renal Agenesis
0008064	Ichthyosis	0001713	Abnormality of Cardiac Ventricle	_	0000085	Horseshoe Kidney
0000988	Skin Rash	0001636	Tetralogy of Fallot	_		·
0001581	Recurrent Skin Infections	0001680	Coarctation of Aorta	_	0000069	Abnormality of the Ureter
0005306	Capillary Hemangiomas	0001647	Bicuspid Aortic Valve	_	0000795	Abnormality of the Urethra
0001597	Abnormality of the Nail Generalized Hypertrichosis	0002616	Aortic Root Dilatation		0000047	Hypospadias
0004554	Alopecia				0000028	Cryptorchidism
0001370	Coarse Hair	0001638	Cardiomyopathy		0000035	Abnormality of the Testis
0002299	Brittle Hair	0011675	Arrhythmia		0000062	Ambiguous Genitalia
				🗆		
RESPIRATOR	γ	METABOLIC		···· MUS	CULOSK	ELETAL
0002093	Respiratory Insufficiency	0001946	Ketosis		0011398	Hypotonia
0002878	Respiratory Failure	0003074	Hyperglycemia		0001276	Hypertonia
0002104	Apnea	0001943	Hypoglycemia		0000098	Tall Stature
0002791	Hypoventilation	0001941	Acidosis		0004322	Short Stature
_					0001382	Joint Hypermobility
0002883	Hyperventilation Recurrent Upper Respiratory Tract	0003128	Lactic Acidosis		0001371	Flexion Contracture
0002788	Infections	0003215	Dicarboxylic Aciduria		0002804	Arthrogryposis Multiplex Congenita
Ш		0002490	Increased CSF lactate		0001161	Hand Polydactly
		0001992	Organic Aciduria		0001829	Foot Polydactly
		0030085	Abnormal CSF Lactate Level	=	0006101	Finger Syndactly
		00003542	2 Increased Serum Pyruvate	_	0001770	Toe Syndactly
GASTROINTE	STINAL	0003535	3-Methylglutaconic aciduria	=	0100490	Camptodactyly of Finger
0002021	Pyloric Stenosis	0001942	Metabolic acidosis	=	0012165	Oligodactyly
0002575	Tracheoesophogeal Fistula	0100493	Hypoammonemia	=	0001762	Talipes Equinovarus
0002032	Esophageal Atresia				0002757	Recurrent Fractures
0002020	Gastroesophageal Reflux	0001987	Hyperammonemia	_	0002650	Scoliosis
0001733	Pancreatitis	0004923	Hyperphenylalaninemia		0002808	Kyphosis
0002014	Diarrhea	0003234	Decreased Plasma Carnitine	_	0003307	Hyperlordosis
0002017	Constipation Inflammatory Bowel Disease	0003236	Elevated Serum Creatine Phosphokinase	=	0001528	Hemihypertrophy
0002037	Intestinal Pseudo-Obstruction	Abnorma	l Newborn Screen	=	0001513	Obesity
0001399	Hepatic Failure	Unusual (	Color/Odor	_	0001548 0002652	Overgrowth Skeletal Dysplasia
0002572	Episodic Vomiting	П			0002032	Svererar nyshrasia
0001744	Splenomegaly			_		
0002240	Hepatomegaly	<u> </u>		⊔_		
0001508	Postnatal Failure to Thrive					
0002578	Gastroparesis					



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## **SEQUENTIAL TRIO WHOLE EXOME SEQUENCING REQUISITION (TEST CODE: 1601)**

		//	
Patient First Name	MI	Date of Birth (MM / DD / YYYY)	Genetic Sex
CONTINUED			
HEMATOLOGY	,	OTHER	
Chr   Cyc   Cyc   O001873   O005537   O005518   O004444   O012410	25549 Congenital ronic clic Thrombocytopenia Macrothrombocytopenia Decreased Mean Platelet Vo Erythrocyte Macrocytosis Spherocytosis Pure Red Cell Aplasia	0001954   Episodic I   0004313   Hypogam   0010701   Abnorma   0002721   Immunod   0012088   Abnorma   0012537   Food into   0008067   Abnorma	maglobulinemia I Immunoglobulins eficiency I urinary odor Ierance Ily lax or hyperextensible skin
Hypin   Hypi	poplastic Anemia Bone Marrow Hypocellularit  ncer gnosis tory of Cancer and Affected R	Family History of Sim  0001254 Lethargy  0002415 Leukodys  GENES OF INTEREST  elatives	ilar Disorder
	0001875	HEMATOLOGY    0001875 Neutropenia   0005549 Congenital   Chronic   Cyclic     0001873 Thrombocytopenia   0005537 Decreased Mean Platelet Vo   0005518 Erythrocyte Macrocytosis   0004444 Spherocytosis   0012410 Pure Red Cell Aplasia   Aplastic   Hypoplastic   Hypoplastic   0001903 Anemia   0005528 Bone Marrow Hypocellularit   Dentation   Age of Diagnosis   Family History of Cancer and Affected R	HEMATOLOGY OTHER    0001875 Neutropenia   Organomegaly     0005549 Congenital   Chronic Infections     Chronic   0004311 Abnormal     Cyclic   0001954 Episodic I     India   0001873 Thrombocytopenia   0010701 Abnormal     0005537 Decreased Mean Platelet Volume   0002721 Immunod     0005518 Erythrocyte Macrocytosis   0012088 Abnormal     0004444 Spherocytosis   0012537 Food intol     0012410 Pure Red Cell Aplasia   0008067 Abnormal     Aplastic   Abnormal Movements     Hypoplastic   Family History of Sim     irrment   0001903 Anemia   0001254 Lethargy     0005528 Bone Marrow Hypocellularity   0002415 Leukodys     nent   Type of Cancer     Type of Cancer   Type of Cancer

Consent on next page



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in X ⊙ (f) **•** 

#### INFORMED CONSENT FOR SEQUENTIAL TRIO WHOLE EXOME SEQUENCING (TEST CODE: 1601) TESTING

			/ /	
Patient Last Name	Patient First Name	MI	Date of Birth (MM / DD / YYYY)	Genetic Sex

#### WHOLE EXOME SEQUENCING (WES) AND WHOLE GENOME SEQUENCING (WGS) CONSENT

This consent form can only be used for whole exome sequencing and whole genome sequencing. Consent forms for other tests are located at Baylor Genetics' website (https://www.baylorgenetics.com/consent/).

For the purposes of this consent, "I", "my", "you", and "your" can refer to you, your child, your unborn child, or other individual you are the legal representative of.

#### **TEST INFORMATION**

Your healthcare provider (doctor, genetic counselor, or other person with medical training) wants to order a genetic test called Whole Genome Sequencing (WGS) or Whole Exome Sequencing (WES). These tests look for changes, called variants, in a person's DNA that can cause health issues. DNA is our genetic material. These variants can be in certain genes, specific parts of our DNA that are needed for our health. They can also be found in other places in the genome (all DNA that a person has). Based on your known health issues, variants in your DNA that may cause these issues will be reported. This test may explain your health issues. It may also explain health issues that your family may have. Even if this test finds the cause of your health issues, this may not help treat or manage those issues.

Testing where your DNA is compared to one or more family members may be performed. This may help better understand your results or show if your family members have the

Before you sign this consent form, you should speak with your healthcare provider. They can help you understand this testing and what it means for your health.

#### **TEST RESULTS**

There are several types of test results that may be reported including:

- Positive: A variant in the DNA was found that is related to your health issues or a health issue that you are at an increased risk of having in the future. These changes that cause disease are also known as pathogenic variants.
- Negative: No variants in the DNA were found that are related to your health issues or that would increase your risk of a health issue in the future.
- Variant of Uncertain Clinical Significance (VUS): A variant in the DNA was found that we do not know its effect, if any, on health. More testing may be needed for you or your family if a VUS is found that may be associated with your health issues.
- Secondary and Incidental Findings (Optional): Testing can sometimes find a variant in the DNA not related to the reason for testing but can change your medical care. Note: Certain issues within the brain start in adulthood and get worse over time (neurodegenerative). They often have no cure or treatment. By default, these variants will not be reported unless they are related to your health issues. However, variants in one or more of these gene(s) can be requested if needed. Your provider must write each gene needed in your test order.
- Genes of No Known Disease Association (Optional): Testing may find a variant in a gene that is not known to cause disease. This may be helpful to learn more about these genes in the future. These results do not currently impact medical management or indicate a diagnosis.

#### SECONDARY AND INCIDENTAL FINDINGS

The following categories of variants are not expected to cause your current health issues. However, they can each be requested to be reported. Knowing about these variants might affect your future medical care.

- ACMG Secondary Findings: The American College of Medical Genetics and Genomics (ACMG) recommends reporting disease-causing variants in certain genes that cause health issues. Each family member can request this group of variants to be reported.
- Incidental Findings: Other variants known to cause health issues but that are not causing your current health issues.

### **CONSIDERATIONS AND LIMITATIONS**

- You should speak with your provider before signing this consent form to understand the risks, benefits, and alternatives to testing.
- Testing may show you have, or are at increased chance of having, a health issue. It may show that you have an increased chance of having a child with a health issue.
- Even if the variant(s) causing your health issues are found, how these issues might progress or improve with treatment might not be known. Affected family members with the same variant might not be affected like you are.
- Depending on the results of testing, more testing may be needed to understand these results. This testing might be needed for you and/or other family members.
- A negative result does not rule out the chance for health issues. Our knowledge of variants and how they cause disease may change over time as we learn more about genetics. Testing has limitations to what it can find as well.
- Certain factors may lead to incorrect results. These include mislabeled samples, incorrect information in the test order, and rare technical errors.
- More sample may be needed from you if the first sample is not sufficient to complete testing

#### FOR SAMPLES FROM NEW YORK STATE RESIDENTS

Samples from New York State residents shall not be included in research without written consent. Samples will not be retained for more than sixty (60) days after receipt by Baylor Genetics, unless authorized by marking below. No tests other than those authorized shall be performed on the samples.

I authorize Baylor Genetics to retain sample(s) longer based on our retention policy for test development, quality assurance, and training purposes.

#### PATIENT CONFIDENTIALITY AND SAMPLE RETENTION

- If several family members are tested, knowing the correct biological relationships among them is important. In rare cases, testing can show that family members are not related as expected. If this is found, we may contact the provider who ordered your testing
- If this testing is requested to be cancelled after the order and sample are sent to the laboratory, please see our Test Cancellation Policy at www.baylorgenetics.com/ cancel-test/.



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## INFORMED CONSENT FOR SEQUENTIAL TRIO WHOLE EXOME SEQUENCING (TEST CODE: 1601) TESTING

			//	
Patient Last Name	Patient First Name	МІ	Date of Birth (MM / DD / YYYY)	Genetic Sex

#### PATIENT CONFIDENTIALITY AND SAMPLE RETENTION (CONTINUED)

- Only Baylor Genetics and its contracted partners will have access to your sample for the ordered testing. Results from testing will only be released to: (i) a licensed healthcare provider, (ii) those authorized in writing, (iii) the patient or their personal representative, and (iv) those allowed access to test results by law. You have the right to access your test results from Baylor Genetics by providing a written request. You also have the right to request raw data obtained from your sample by providing a written request or HIPAA Authorization Form.
- In rare cases, people with genetic diseases may have problems with health insurance and employment. The U.S. Federal Government has several laws that prohibit discrimination based on test results by health insurance companies and employers. These laws also prohibit unauthorized disclosure of this information. For more information, please visit www.genome.gov/10002077.
- Samples will be kept in the laboratory based on our retention policy. Once testing is completed, the de-identified sample may be used for test development, quality assurance, and training purposes. Samples are not returned to patients or providers unless requested prior to testing. You and your heirs will not receive payments, benefits, or rights to any resulting products or discoveries.
- The information from your testing may be used in scientific research, publications or presentations, but your specific identity will not be revealed. We may contact your provider to obtain more clinical information about you. Baylor Genetics also performs other types of scientific research and may contact you to see if you would like to be involved.
- Variants found may be submitted to databases. The medical community uses these databases to collect information about how variants might cause disease to improve testing and treatment for patients. An example is ClinVar, a free, public archive of reports on human genetics. Limited clinical information may need to be shared with these databases. In rare cases, this information may be enough to allow you or your family members to be identified.
- For more information on privacy practices at Baylor Genetics, please visit www.baylorgenetics.com/privacy-practices/.

#### FINANCIAL AGREEMENT

By signing below, I hereby authorize Baylor Genetics to provide my insurance carrier any information necessary, including test results, for processing my insurance claim. I understand that I am responsible for any co-pay, co-insurance, and unmet deductible that the insurance policy dictates. I designate Baylor Genetics as my designated representative for purposes of appealing any denial of benefits by my insurance carrier. I irrevocably assign associated payment to Baylor Genetics, and direct that payment be made directly to Baylor Genetics. Please note, some payers may not cover certain screening tests.

If my health insurer does not cover the test or I do not have health insurance, I have received a good faith estimate of the cost for the genetic testing ordered by my provider and agree to pay for the cost of the genetic testing billed to me by Baylor Genetics based on that good faith estimate. More information is available in Baylor Genetics' No Surprises Act and Good Faith Estimate Notice located at https://www.baylorgenetics.com/no-surprises-act/.

A Medicare Advance Beneficiary Notice (ABN) is required for services Medicare identifies as not medically necessary.

#### PATIENT AUTHORIZATION

By signing this statement of consent, I acknowledge that I have read, understand, and hereby grant my informed consent for genetic testing. I have received appropriate explanations from my healthcare provider about the planned genetic test(s) and possible results. I have been informed by my healthcare provider about the availability and importance of genetic counseling and have been provided with written information identifying a genetic counselor or medical geneticist who can provide such counseling services. All my questions have been answered, and I have had the necessary time to make an informed decision about the genetic test(s).

Note: If Prenatal WES was ordered, please leave the Patient section blank and complete only a section for each relative tested below. I hereby give permission to Baylor Genetics to conduct genetic testing as recommended by my healthcare provider.\* Date Signed (MM / DD / YYYY) Patient Name Patient Signature Relationship to Patient Name Signature Date Relative 1 Relative 2 Relative 3 If one or more family members have a Representative signing on their behalf: Date (MM / DD / YYYY) Name Signature Representative For Relationship to Represented Person(s) \*If you are signing on behalf of the patient as the parent(s) and/or person with legal authority to act on behalf of the patient or parent, you may be required to provide evidence of your authority. FOR SURROGATES PREGNANCIES - FOR PRENATAL WES ONLY:

Maternal cell contamination (MCC) studies use blood or another sample from a pregnant person. MCC studies are used to determine that the sample being tested belongs to the fetus and not the pregnant person. The results of MCC studies are not used for the treatment or management of the fetus, pregnant person, or other individuals, and are not part of the pregnant person's designated medical record.

I hereby give permission for my sample to be used for MCC studies:

Surrogate Name Surrogate Signature