PREVENTION GENETICS

PREVENTIONGENETICS USE ONLY

Test information is available on our website:
www.PreventionGenetics.com

All testing must be ordered by a qualified Healthcare Provider

THIS FORM MUST ACCOMPANY ALL SPECIMENS

TEST REQUISITION FORM
SP051 - Inozyme Pharma

ALL FIELDS REQUIRED

WHEN TESTING A PREGNANCY - SKIP PAGE 1 AND COMPLETE PAGES 2-4

PERSON COMPLETING FORM	CONTACT (PHONE OR EMAIL)	DATE OF REQUEST
		______ / ______ / ______ MONTH DAY YEAR

PATIENT INFORMATION

LAST (FAMILY) NAME	FIRST NAME	MI	DATE OF BIRTH
			______ / ______ / ______ MONTH DAY YEAR

PATIENT ID

SPECIMEN COLLECTION DATE
MONTH ______ / DAY ______ / YEAR ______

SEX
☐ Male ☐ Female
☐ Other
______ SPECIFY KARYOTYPE

HAS PATIENT BEEN TESTED PREVIOUSLY AT PreventionGenetics?
☐ NO
☐ YES, PG ID# ______

SPECIMEN SOURCE
☐ Blood ☐ Saliva
☐ Other ______

ANCESTRY

HAS PATIENT'S RELATIVE BEEN TESTED PREVIOUSLY AT PreventionGenetics?
☐ NO ☐ YES
Name ______
DOB ______ / ______ / ______ (MONTH DAY YEAR) Relationship ______
or PG ID# ______

REASON FOR TEST
☐ Diagnosis / Affected
☐ Presymptomatic / At Risk
☐ Carrier Testing

BLOOD TRANSFUSION
☐ NO
☐ Within Last 30 Days, Date and Type
______ / ______ / ______ MONTH DAY YEAR
TYPE ______

BONE MARROW TRANSPLANT
☐ NO
☐ YES
______ / ______ / ______ MONTH DAY YEAR

OTHER RELEVANT CLINICAL INFORMATION (Labs, biopsies, other genetic testing performed, etc.) ***PLEASE ATTACH PEDIGREE, IF POSSIBLE.***

TEST SELECTION

TAT begins from date PreventionGenetics receives specimen and signed Inozyme Pharma informed consent form.

TEST CODE	DESCRIPTION	TAT	ADDITIONAL INFORMATION
☐ 7555	***ENPP1* and *ABCC6* Sequencing Includes Deletion / Duplication Testing**	14 Days	*Appropriate for individuals / deceased individuals with a clinical suspicion of GACI or ARHR2, who meet eligibility criteria (see informed consent form).*
☐ 100	**Testing for One Variant** Previously identified in the family	14 Days	LIST THE GENE / VARIANT TO BE TESTED
☐ 200	**Testing for Two Variants** Previously identified in the family	14 Days	LIST THE TWO GENES / VARIANTS TO BE TESTED

COMMENTS

SPECIAL INSTRUCTIONS

SP051

☐ **POSITIVE CONTROL**
No report will be issued

☐ **SPECIMEN COLLECTED IN NEW YORK STATE**
Include New York State Non-Permitted Laboratory Test Request approval letter and Genetic Testing Healthcare Provider Statement.

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PRENATAL TESTING SP051 - INOZYME PHARMA

WHEN TESTING A PREGNANCY - SKIP PAGE 1 AND COMPLETE PAGES 2-4

IF TESTING IS NOT RELATED TO PREGNANCY, SKIP PAGES 2 - 3, AND COMPLETE PAGES 1 AND 4.

ORDERING CHECKLIST

- ☐ Fetal Specimen
- ☐ Family member specimen(s) - as needed
- ☐ Informed Consent Form
- ☐ Prenatal Healthcare Provider Statement

INSTRUCTIONS

- Fetal, parental and/or proband information must be completed on one form.
- See page 5 for further ordering details.

PERSON COMPLETING FORM	CONTACT (PHONE OR EMAIL)	DATE OF REQUEST
		____ / ____ / ____ (MONTH / DAY / YEAR)

FETAL AND MATERNAL INFORMATION

LAST (FAMILY) NAME

MOTHER'S FIRST NAME (FETUS OF)

MI

MOTHER'S DATE OF BIRTH ____ / ____ / ____ (MONTH / DAY / YEAR)

MATERNAL ID CODE

FETAL SAMPLE COLLECTION DATE ________ TIME ☐ AM ☐ PM ____ / ____ / ____ (MONTH / DAY / YEAR)

FETAL SEX
☐ Male ☐ Female
☐ Unknown ☐ Ambiguous
Based On: ______________

PRENATAL SPECIMEN SOURCE

- ☐ Cell Culture, Source ______________
- ☐ Direct Amniotic Fluid
- ☐ Fetal Blood (PUBS)
- ☐ Extracted DNA, Source ______________
- ☐ Direct CVS
- ☐ Other, Source ______________

WILL A BACKUP SAMPLE BE MAINTAINED AT ANOTHER LOCATION?
☐ NO ☐ YES

ADDITIONAL MATERNAL INFORMATION

MATERNAL SPECIMEN SOURCE

- ☐ Whole Blood 5mL EDTA - Preferred
- ☐ Extracted DNA, Source ______________
- ☐ Saliva
- ☐ Other, Source ______________

DATE COLLECTED MONTH ________ / DAY ________ / YEAR ________

CLINICAL FEATURES
☐ Unaffected ☐ Unknown ☐ Affected, features ______________

ANCESTRY

HAS PATIENT BEEN TESTED PREVIOUSLY AT PreventionGenetics?
☐ NO ☐ YES, PG ID# ______________

BLOOD TRANSFUSION
☐ NO
☐ Within Last 30 Days, Date ____ / ____ / ____ (MONTH / DAY / YEAR)

BONE MARROW TRANSPLANT
☐ NO
☐ YES ____ / ____ / ____ (MONTH / DAY / YEAR)

PREGNANCY HISTORY

GESTATIONAL AGE AT SAMPLE COLLECTION ________ ☐ by U/S ☐ by LMP

IS THIS AN ONGOING PREGNANCY? ☐ No ☐ Yes

DONOR PREGNANCY ☐ No ☐ Yes

MULTIPLE GESTATION PREGNANCY? ☐ Twins ☐ Triplets ☐ Other ________

PATERNAL INFORMATION (Targeted Prenatal Testing Only, if needed)

LAST (FAMILY) NAME

FIRST NAME

MI

DATE OF BIRTH ____ / ____ / ____ (MONTH / DAY / YEAR)

PATERNAL SPECIMEN SOURCE

- ☐ Whole Blood 5mL EDTA - Preferred
- ☐ Extracted DNA, Source ______________
- ☐ Saliva
- ☐ Other, Source ______________

DATE COLLECTED ____ / ____ / ____ (MONTH / DAY / YEAR)

PATIENT ID CODE

CLINICAL FEATURES
☐ Unaffected ☐ Unknown ☐ Affected, features ______________

ANCESTRY

HAS PATIENT BEEN TESTED PREVIOUSLY AT PreventionGenetics?
☐ NO ☐ YES, PG ID# ______________

BLOOD TRANSFUSION
☐ NO
☐ Within Last 30 Days, Date ____ / ____ / ____ (MONTH / DAY / YEAR)

BONE MARROW TRANSPLANT
☐ NO
☐ YES ____ / ____ / ____ (MONTH / DAY / YEAR)

PREVENTION GENETICS

PREVENTIONGENETICS USE ONLY

THIS FORM MUST ACCOMPANY ALL SPECIMENS

ADDITIONAL FAMILY MEMBER INFORMATION (Targeted Prenatal Testing Only, if needed)

LAST (FAMILY) NAME | FIRST NAME | MI | DATE OF BIRTH ____ / ____ / ____ (MONTH / DAY / YEAR)

SPECIMEN SOURCE
- ☐ Whole Blood 5mL EDTA - Preferred
- ☐ Extracted DNA, Source ______________
- ☐ Saliva
- ☐ Other, Source ______________

DATE COLLECTED ____ / ____ / ____ (MONTH / DAY / YEAR)

PATIENT ID CODE

CLINICAL FEATURES
☐ Unaffected ☐ Unknown ☐ Affected, features ______________

ANCESTRY

BLOOD TRANSFUSION
☐ NO
☐ Within Last 30 Days, Date ____ / ____ / ____ (MONTH / DAY / YEAR)

BONE MARROW TRANSPLANT
☐ NO
☐ YES ____ / ____ / ____ (MONTH / DAY / YEAR)

HAS PATIENT BEEN TESTED PREVIOUSLY AT PreventionGenetics?
☐ NO ☐ YES, PG ID# ______________

SEX
☐ Male ☐ Female ☐ Unknown/Other

PRENATAL TESTING - TEST SELECTION

FETAL TEST SELECTION

Check the box next to the test that is to be performed. If targeted testing, please include details. All testing related to an ongoing pregnancy is courtesy expedited.
We require at least one parental specimen be sent for prenatal testing. See page 5 for more information.

TEST CODE	TEST NAME	GENE(S)	VARIANT(S)
☐ 990	Targeted Prenatal Testing for Known Familial Variants. Includes STAT turnaround time (8-10 calendar days); positive control required.		
☐ 7555	*ENPP1* and *ABCC6* Sequencing and Deletion/Duplication Testing 14 day TAT	SPECIAL INSTRUCTIONS: ☐ SPECIMEN COLLECTED IN NEW YORK STATE Include New York State Non-Permitted Laboratory Test Request approval letter and Genetic Testing Healthcare Provider Statement.	

MATERNAL TEST SELECTION

For Targeted Prenatal Testing (Test Code 990), positive controls from parents and/or proband are required. Maternal Cell Contamination (MCC) Studies (Test Code 800) are strongly recommended for any fetal testing.

TEST	GENE(S)	VARIANT(S)
☐ Positive Control for Variant(s) Test Code 100, 200, or 300		
☐ Maternal Cell Contamination (MCC) Study Test Code 800		

PATERNAL TEST SELECTION

For Targeted Prenatal Testing (Test Code 990), positive controls from parents and/or proband are required.

TEST	GENE(S)	VARIANT(S)
☐ Positive Control for Variant(s) Test Code 100, 200, or 300		

ADDITIONAL FAMILY MEMBER TEST SELECTION

For Targeted Prenatal Testing (Test Code 990), positive controls from parents and/or proband are required.

TEST	GENE(S)	VARIANT(S)
☐ Positive Control for Variant(s) Test Code 100, 200, or 300		

ADDITIONAL CLINICAL INFORMATION *(STRONGLY RECOMMENDED)*

Other relevant clinical information (labs, ultrasound results, biopsies, other genetic testing performed, etc. Please attach a pedigree, if available.

PREVENTION GENETICS

PREVENTIONGENETICS USE ONLY

THIS FORM MUST ACCOMPANY ALL SPECIMENS

PROVIDER / LABORATORY CONTACT INFORMATION

Our preferred method of report transmission is secure email (via ZixCorp).
Please provide an email address when possible. If you have additional specific reporting requests, indicate them below.

PROVIDER INFORMATION

INSTITUTION

ADDRESS (City, State, Country and Postal Code)

REQUESTING PHYSICIAN (First, Last, Degree)		REQUESTING GENETIC COUNSELOR OR ALLIED PROVIDER (First, Last, Degree)	
PHONE NUMBER	NPI#	PHONE NUMBER	NPI#
EMAIL		EMAIL	

TEST REPORTING INSTRUCTIONS
Our preferred method of report transmission is email via ZixCorp

SECURE EMAIL VIA ZIXCORP ☐ *Use above email address*

☐ DO NOT USE ZIXCORP. EMAIL RESULTS VIA SHAREFILE.
☐ DO NOT EMAIL RESULTS. Send via fax (provide fax number):

(________) ______-__________

TEST REPORTING INSTRUCTIONS
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☐ DO NOT EMAIL RESULTS. Send via fax (provide fax number):

(________) ______-__________

SENDOUT LABORATORY COMPLETE ONLY IF REPORT IS NEEDED

INSTITUTION / CONTACT

ADDRESS (City, State, Country and Postal Code)

PHONE NUMBER	NPI# (Where Applicable)

EMAIL

TEST REPORTING INSTRUCTIONS
Our preferred method of report transmission is email via ZixCorp

SECURE EMAIL VIA ZIXCORP ☐ *Use above email address*

☐ DO NOT USE ZIXCORP. EMAIL RESULTS VIA SHAREFILE.
☐ DO NOT EMAIL RESULTS. Send via fax (provide fax number):

(________) ______-__________

OTHER

INSTITUTION / CONTACT

ADDRESS (City, State, Country and Postal Code)

PHONE NUMBER	NPI# (Where Applicable)

EMAIL

TEST REPORTING INSTRUCTIONS
Our preferred method of report transmission is email via ZixCorp

SECURE EMAIL VIA ZIXCORP ☐ *Use above email address*

☐ DO NOT USE ZIXCORP. EMAIL RESULTS VIA SHAREFILE.
☐ DO NOT EMAIL RESULTS. Send via fax (provide fax number):

(________) ______-__________

INSTITUTIONAL BILLING

BILLING INSTITUTION	Inozyme Pharma	CUSTOMER NUMBER	INOZYME 10010	SPECIAL PROJECT NUMBER	SP051

PREFERRED SPECIMEN REQUIREMENTS

PLEASE CONTACT US WITH ADDITIONAL SPECIMEN REQUIREMENT QUESTIONS.

WHOLE BLOOD

Collect 3 ml - 5 ml of whole blood in EDTA (purple top tube) or ACD (yellow top tube), minimum 1 ml for infants 6 months of age or less.

DNA

Send in screw cap tube at least 5 µg -10 µg of purified DNA at a concentration of at least 20 ng/µL. Indicate concentration on tube label.

SALIVA

Oragene™ or GeneFiX™ Saliva Collection kit used according to manufacturer instructions.

FRESH, FROZEN TISSUE

Collect 2mm x 2mm x 2mm tissue and flash freeze. Tissue to be sent frozen (preferably on dry ice). Contact us for additional details.

FETAL (CVS / AMNIOCYTES) AND OTHER CELL CULTURES

Culture and send at least two, T25 flasks of confluent cells. Multiple test requests may also require additional flasks. Please contact us for details. We strongly recommend maintaining a back-up culture.

DIRECT AMNIOTIC FLUID / CHORIONIC VILLI

Collect 10 ml -20 ml of direct amniotic fluid or 5 mg -10 mg cleaned CVS tissue (about 15-20 cleaned villi). We strongly recommend maintaining a local back-up culture. Direct Amniotic Fluid and Villi samples are accepted only for targeted variant testing.

PRENATAL INSTRUCTIONS

PreventionGenetics should be notified in advance of arrival of a prenatal specimen. For all prenatal testing in ongoing pregnancies, we require a signature from the health care provider on the Inozyme Pharma Informed Consent Form. We expect that the ordering provider will take responsibility for the appropriateness of the requested testing.

We accept fetal DNA, fetal tissue, cultured fetal cells, or direct CVS / amniotic fluid. However, acceptable specimen type is dependent on the fetal testing requested. Direct CVS or amniotic fluid samples can be accepted only for targeted prenatal testing. Any of the aforementioned sample types are accepted for full gene sequencing. Retention of a backup culture of the fetal cells is strongly recommended. Where possible, please ship prenatal samples so that they will arrive at PreventionGenetics no later than Friday.

We require at least one parental specimen be sent as part of prenatal testing for QA purposes. If targeted prenatal testing is ordered, we must receive a positive control sample(s) also.

Maternal cell contamination (MCC) of fetal sample will be tested using the PreventionGenetics DNA Genotyping Panel. There is not a separate report for MCC studies.

PreventionGenetics does not perform prenatal testing for sex. We will also not report fetal sex unless this is critical for interpretation of test results. PreventionGenetics does not perform pre-implantation DNA testing.

FAMILIAL VARIANT TESTING FOR PREGNANCY

TEST CODE 990

Familial variants must be known in advance from testing of parents, affected siblings or other relatives. These variants must be confirmed at PreventionGenetics in the parents and/or proband. Parental and positive control specimens may be sent in advance of the prenatal specimen. We require at least one parental specimen be sent for all targeted prenatal testing requests. Please order Test #100 or Test #200 as appropriate for positive control and parental samples on their own test requisition forms. An informed consent form is not required for positive control samples. Turnaround Time: 8-10 calendar days from receipt of specimen and signed Inozyme Pharma Informed Consent Form.

NEXT GENERATION SEQUENCING FOR PREGNANCY

TEST CODE 7555

We require at least one parental specimen be sent for all prenatal testing requests. Test code 7555 includes deletion/duplication testing via Copy Number Variant detection from NextGen data. Turnaround Time: The great majority of NGS tests are completed within 14 days from date of specimen and signed Inozyme Pharma Informed Consent Form receipt.

FETAL CELL CULTURE

We do not culture cells at PreventionGenetics. We strongly encourage you to maintain a local back up culture. If we are unable to complete testing with the sample that was provided, we will reach out to the provider to request the back up culture.

SHIPPING AND HANDLING INSTRUCTIONS

Please label all specimen containers with the patient's name, date of birth and/or ID number. At least two identifiers should be listed on specimen containers. We accept specimen deliveries Monday-Saturday for all specimen types except cell cultures, direct amniotic fluid, or direct chorionic villi. Cell culture deliveries are routinely accepted Monday-Friday and require advance notice of arrival. If a Saturday delivery is necessary, please contact us to make arrangements. Saturday delivery should especially be avoided when possible as prenatal specimens are not processed over the weekend. Holiday schedules will be posted on our home page at least one week prior to major holidays.

BLOOD

DO NOT FREEZE. During hot weather, include a frozen ice pack in the shipping container. Place a paper towel or other thin material between the ice pack and the blood tube. In cold weather include an unfrozen ice pack in the shipping container as insulation. At room temperature, a blood specimen is stable for up to 48 hours. If refrigerated, a blood specimen is stable for up to one week.

DNA

DNA may be shipped at room temperature. Label the tube with the composition of the solute, DNA concentration as well as the patient's name, date of birth, and/or ID number. We only accept genomic DNA for testing. We do not accept products of whole genome amplification reactions or other amplification reactions.

PRENATAL TESTING

Please contact us in advance regarding prenatal test requests. When possible, ship prenatal samples to arrive at PreventionGenetics no later than Friday.

CELL CULTURES, DIRECT AF/CVS, AND POC

Send specimens overnight in an insulated, shatterproof container. Direct AF/CVS or POC specimens can be sent in saline or culture media at room temperature.

DNA GENOTYPING PANEL

For quality control purposes, the PreventionGenetics DNA Genotyping Panel is performed on all clinical specimens. Genotyping results are not included in test reports.

DNA BANKING

DNA Banking is available, but is outside the scope of this program. Visit our website at www.PGDNABank.com for information about the process and forms. For questions related to PGDNABanking, contact our DNA Banking Director at (715) 387-0484, ext. 151, or email: dnabanking@preventiongenetics.com.

CONTACT US

For additional questions or concerns, please contact our Client Service Representatives at (715) 387-0484, ext. 0, or our Genetic Counseling Team at option 2, or email: clinicaldnatesting@preventiongenetics.com.

ADDRESS

PreventionGenetics - Diagnostic Lab
3800 S. Business Park Ave.
Marshfield, Wisconsin 54449
USA

TESTING KITS

Clinical testing kits with prepaid return shipping are available. To order test kits, submit requests through our electronic order form (see website) or contact our Client Service Representatives at (715) 387-0484, ext. 0.

COMMENT SP051 when ordering kits.