PerkinElmer For the Better | PerkinElmer GENOMICS

Agios Clinical Genomics Requisition Form

This requisition form can be used to submit a specimen for the Anemia Identified program, a no-charge US testing program sponsored by Agios and offered through Perkin Elmer. All patients suspected of having, or with a family history of, Hereditary Anemia (HA) can take part in the Anemia Identified Program.

The Anemia Identified program facilitates access to genetic testing to help in the diagnosis of HA or carrier status identification of HA. While Agios provides financial support for this program, tests and services are performed by an independent third party, PerkinElmer. Healthcare providers must confirm that patients meet certain criteria to use the program. Agios receives de-identified patient data from this program, but at no time does Agios receive patient identifiable information. Agios receives contact information for healthcare providers who use this program. Genetic testing is available in the U.S. and Puerto Rico only. Healthcare providers who use this program have no obligation to recommend, purchase, order, prescribe, promote, administer, use or support any Agios product.

Please complete every field and tick box clearly.

PATIENT INFORMATION

Patient's First Name | Middle Initial | Patient's Date of Birth (MM/ DD /YYYY)

Patient's Last Name | Patient ID Number

Biological Sex: ○ Male ○ Female ○ Unknown

Patient's Street Address

City / Town | State | Zip Code

Country | Patient's Preferred Phone

Patient's Email

Ethnicity (check all that apply):	○ African-American	○ Asian (China, Japan, Korea)
○ Jewish - Ashkenazi	○ Caucasian/N. European/S. European	
○ Hispanic	○ Middle Eastern (Saudi Arabia, Qatar, Iraq, Turkey)	
○ Native American	○ Other (specify):_____	

ORDERING PROVIDER

Provider's First and Last Name

Provider Account Number | NPI

Clinic/Hospital/Institution Name

Provider's Email | Provider's Phone

Provider's Street Address

City / Town | State | Zip Code

Country | Provider's Fax

SEND ADDITIONAL COPY OF RESULTS TO (If applicable)

Name

Provider/Contact Account # | Phone Number

Email Address | Fax Number

PATIENT SAMPLE INFORMATION

SAMPLE TYPE:
○ Saliva Swab
○ Whole Blood

Collection Date: MM/DD/YY

Was this sample collected in NY State: ○ yes ○ no

INDICATION FOR TESTING

○ Diagnosis in symptomatic patient
○ Carrier testing
○ Presymptomatic testing of at-risk family member
○ Other:_____

TEST MENU

○ AGS001 Hereditary Anemia Panel
○ AGS002 Pyruvate Kinase Enzyme Activity

X INSTITUTIONAL BILLING

Agios | Institution/Organization Name

B0374 | Institution/Organization Account #

Do not bill patient's insurance.

Test Ordered	Acceptable Sample	Cold Pack Needed?
Hereditary Anemia Panel	1 Whole Blood (EDTA) sample or 1 saliva sample	NO
Pyruvate Kinase Enzyme Activity	1 Whole Blood (EDTA) sample	YES*
Hereditary Anemia Panel and Pyruvate Kinase Enzyme Activity together	2 Whole Blood (EDTA) samples	YES*

* Follow the instructions included in the whole blood collection pack to ensure proper usage of the cold pack.

PHYSICIAN CONFIRMATION OF INFORMED CONSENT AND MEDICAL NECESSITY

The undersigned person (or representative thereof) ensures he/she is a licensed medical professional authorized to order genetic testing and confirms that the patient has given appropriate informed consent for the testing ordered, including a discussion of the benefits and limitations. I confirm that testing is medically necessary and that test results may impact medical management for the patient. Furthermore, all information on this TRF is true to the best of my knowledge. My signature applies to the informed consent and/or attached letter of medical necessity.

I consent to PerkinElmer sharing my name and contact information, and the date on which I executed this TRF, with Agios Pharmaceuticals and I understand and agree that I may be contacted by Agios Pharmaceuticals in connection with the genetic testing program for the purpose of receiving information on hereditary anemias, including pyruvate kinase deficiency.

Signature_____ Date_____

FOR INTERNAL USE ONLY

Date Rec'd_____ Rec'd _____

TEMP	SPEC	COL	#TUBES	VOL
R/C/F				
R/C/F				
R/C/F				

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ADDITIONAL OPTIONAL PHENOTYPE / PATIENT HISTORY SECTION

Clinical History

CBC Data

WBC: _____
HGB: _____
HCT: _____
RBC: _____
MCV: _____
MCH: _____
CHC: _____
RDW: _____
PLT: _____
Rectics %: _____
Abs Retic: _____
Ferritin: _____

Relevant Clinical Information

○ Asymptomatic ○ Symptomatic: _____
○ Acquired ○ Lifelong/familial ○ Perinatal/neonatal ○ Chronic ○ Episodic/sporadic
Recent transfusion: ○ Yes ○ No Last transfusion date *(mm-dd-yyyy)*: _____
Family history: ○ Yes ○ No Disorder/relation to patient: _____
Parental consanguinuity: ○ Yes ○ No
Blood smear shows: _____
Bone marrow shows: _____

Indication for Testing (See Metabolic Hematology Profile Comparison Chart)

Suspect

○ Hereditary spherocytosis
○ Hereditary elliptocytosis
○ Hereditary pyropoikilocytosis
○ Hereditary stomatocytosis
○ Pyruvate Kinase Deficiency
○ Southeast Asian ovalocytosis
○ Congenital dyserythropoietic anemia
○ Enzyme disorder: _____
○ Other: _____

Previous Results

Previous protein/functional testing: ○ Yes _____
Hb electrophoresis: _____

○ G6PD activity level: _____ Coombs: ○ Pos ○ Neg ○ Not Done
○ IK activity level: _____ Splenectomy: ○ Yes ○ No
○ Other enzyme level(s): _____
○ Osmotic fragility: ○ Normal ○ Increased ○ Decreased ○ Not performed
○ EMA binding/Band3: ○ Normal ○ Abnormal ○ Not performed
○ Ektacytometry: _____

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AGIOS U.S. CLINICAL INFORMED CONSENT FORM

PerkinElmer Genetics, Inc., ("PerkinElmer") requires a completed Patient's Informed Consent Form (ICF) for testing to be performed. The ICF must be completed by the patient, or a legally authorized representative of the patient (or by the healthcare provider where permitted under applicable law or regulation). For any patient below the age of majority, the ICF must be completed by the patient's legally authorized representative.

The purpose of this ICF is to provide you with a description of the Test ordered, known risks and benefits of the Test, anonymization of personal health information ("PHI"), sample and data retention, research opportunities, and the reporting of secondary findings, if applicable. Given the complexity of the type of the Test, it is recommended that you and/or your child receive genetic counseling by a trained genetics professional before and after the testing is performed. There is no cost to you for the Test(s). Funding is provided by Agios Pharmaceuticals.

TEST INFORMATION

Your healthcare provider ("HCP") has recommended that you, or your child, receive enzymatic, biochemical or molecular genetics clinical testing ("Test") indicated on the submitted Test Requisition Form ("Requisition"). For more information on the reasons your HCP has ordered the Test, and the disorders your HCP is having you tested for, please consult with your HCP. You are free to decide if you want this Test performed or not. Providing a Sample and undergoing the Test is voluntary and you may withdraw your consent without penalty at any time.

Enzyme/Biomarker Test: This type of test measures the presence or absence of enzymes/biomarkers and/or their level of activity in an individual. Only the enzymes/biomarkers identified on the requisition will be tested. Results from this type of Test may indicate the presence of a specific condition or conditions, and follow-up confirmatory testing may be recommended.

Genetic/Genomic Test: This type of Test analyzes one or more segments of your DNA depending on the assay requested. This Test is used to identify what, if any, DNA variant(s) you or your child is carrying which is causing the specific disease or condition you are being tested for. Identifying the mutation may be useful for diagnostic and treatment purposes, and allows at-risk family members to be tested. Only the genes identified on the Requisition will be analyzed. In some cases, we may not be able to determine with certainty which gene is actually causing the disease.

TEST METHOD

If you consent to the Test, your HCP will take a sample of your and/or your child's blood, saliva, body fluid, tissue or other sample type. Your Sample will be sent to PerkinElmer's laboratories in the United States for the Test; the majority of testing will be performed at our laboratory headquarters in Pittsburgh, PA.

Under some circumstances, including inadequate or poor quality sample, an additional Sample may be required for Tests to be performed.

TEST RESULTS

Your treating HCP has sole responsibility for all decisions concerning the possible management of your diagnosis and disease; PerkinElmer will not provide a diagnosis. PerkinElmer will report Test results only to your HCP via secure email, a secure internet portal, or fax. Your HCP is responsible for communicating with you regarding the results of the Test and may refer you or your child to a specialist for further clinical evaluation and confirmation of diagnosis, if applicable. Possible results for Genetic/Genomic Tests include:

1. *Positive:* A positive genetic test result may indicate that you are a carrier of, predisposed to, or have the specific disease or condition being tested for. A positive genetic test may limit your access to health insurance or life assurance coverage; for example, a life insurance company might ask you to provide genetic information indicating a disorder if this information is available to you.
2. *Negative:* A negative result indicates that no disease-causing variant was identified in the Test performed. No Test can rule out all genetic diseases or conditions. A negative result does not guarantee that you are free from genetic disorders or other medical conditions.
3. *Inconclusive/Variant of Uncertain Significance:* A variant of uncertain significance (VOUS) result indicates that a DNA change was detected, but it is currently unknown if the variant is associated with a genetic disorder. A VOUS is not the same as a positive result and does not clarify whether there is an increased risk to develop a genetic disorder. The variant could be a benign change or it could be indicative of disease/disease-causing.
4. *Unexpected Results:* In rare instances, this Test may reveal an important genetic change that is not directly related to the reason for ordering this test. This information would be disclosed to your HCP if it potentially impacts medical care, and you have consented to receive this type of result

TEST REPORT

Reported disease-causing variants are described as pathogenic variant(s), likely pathogenic variants(s), or variant(s) of uncertain significance in genes interpreted to be responsible for, or potentially contributing to, a disease or condition. In addition, variants in genes not known to be associated with disease but for which there is evidence to suggest an association with disease may also be reported. For testing performed on prenatal samples or for screening of apparently healthy individuals, only variants classified as pathogenic or likely pathogenic will be reported.

When Whole Exome Sequencing (WES) or Whole Genome Sequencing (WGS) tests are ordered by your HCP, you have the option to receive some findings not directly related to the reason for ordering the Test called "Secondary Findings". When Secondary Findings are requested, only Pathogenic or Likely Pathogenic findings will be reported, where applicable. Please read the Secondary Findings sections on page 3 and/or 4 of this consent form for more information, and available reporting options. For prenatal samples, secondary findings for the proband are not available.

INFORMATION ABOUT PARENTAL AND FAMILIAL SAMPLES

In some circumstances, it may be helpful for additional family members to undergo testing in order to provide information that can aid in the interpretation of the WES/WGS test results. These Tests could be part of a TRIO Test or as stand-alone targeted testing. PerkinElmer, in consultation with the HCP, will decide if other family members need to be tested. If the HCP recommends testing for additional family members, only the Test performed will be reported. If undergoing a TRIO WES or WGS test, family members will have the option to receive information about secondary findings either as a part of the proband report or as a standalone parental report. A full analysis of the parental samples for secondary findings will only be completed if standalone reports are selected (for an additional charge). If family members elect to receive information about secondary findings either as part of the proband report or as a standalone report, the family member must sign all applicable sections on page 3 and/or 4 of this form.

TEST LIMITATIONS

Due to current limitations in technology and incomplete knowledge of diseases and genes, some variants may not be detected by the Test ordered. There is a possibility that the Test result that is uninterpretable or of unknown significance may require further testing when more information is gained. In rare circumstances, Test results may be suggestive of a condition different from that which was originally considered for the purpose of consenting to this Test. The Test may also find variants or genes that lead to conditions for which you currently do not have symptoms or may not be related to your current condition.

TEST RISKS

Patients and family members may experience anxiety before, during, and/or after testing. Testing multiple family members may reveal that familial relationships are not biologically what they were assumed to be. For example, the Test may indicate non-paternity (the stated father of an individual is not the biological father) or consanguinity (the parents of an individual are closely related by blood). These biological relationships may need to be reported to the HCP who ordered the test.

Taking a blood or tissue sample from you and/or your child may lead to mild pain, bruising, swelling, redness, and a slight risk of infection. Light-headedness, fainting or nausea may occur if your HCP collects blood or tissue samples. These side-effects are typically brief and transient, but you should contact your HCP if you and/or your child require treatment. Under some circumstances an additional sample may be required for Tests to be performed.

A positive test result may limit your access to health insurance or life assurance coverage; for example, a life insurance company might ask you to provide genetic information indicating a disorder if this information is available to you. Please refer to information on the Genetic Information Nondiscrimination Act (GINA) and applicable local laws for more information.

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CONFIDENTIALITY

You have the right to confidential treatment of the Sample and your PHI. Your HCP will provide PerkinElmer with Personal Health Information ("PHI") such as your name, date of birth, gender and clinical symptoms to help track your sample and report results. To maintain confidentiality, the test results will only be released to the referring health care provider, to the ordering laboratory, to the patient/guardian, to other health care providers involved in your diagnosis and treatment, or as otherwise required by law or regulation. Unless required by law, PerkinElmer will not disclose your PHI to any person or entity except with your written consent.

You and your HCP can control how your Sample and PHI are processed. You have the right to request access to your PHI, request corrections of any errors in recorded PHI, or where PHI may be missing or incomplete ask that it be completed. You also have the right to ask that your PHI be erased, subject to law or regulation. You can contact your HCP for such requests and your HCP will contact PerkinElmer, or you can contact PerkinElmer directly by visiting www.perkinelmergenomics.com. If requests for access, correction, completion, or erasure cannot be fulfilled, you will be informed and provided with the reasons why your requests cannot be fulfilled.

SAMPLE AND DATA RETENTION

Pursuant to laboratory best practices, your DNA sample will be retained by PerkinElmer for a minimum of two years and then destroyed. Additionally, your PHI, the data from the Tests (including those performed before any withdrawal of consent) and the related reports will be retained by PerkinElmer indefinitely, unless otherwise noted. In some instances, it may be beneficial to you for PerkinElmer to retain your sample for a longer period of time in order to conduct additional testing, and PerkinElmer will do so with appropriate documentation from you or your HCP.

PerkinElmer is requesting consent to keep you and/or your child's anonymized sample and data indefinitely for ongoing test development, scientific research, and/or other activities. This consent is optional, and the Test will be performed whether or not you provide consent to the following:

- PerkinElmer will anonymize and retain your Sample indefinitely for internal quality control, test validation, assay development and improvement. By allowing PerkinElmer to retain your Sample, you understand and agree that you give up any property rights you may have in the Sample and are donating it to PerkinElmer Genetics, Inc. If you withdraw your consent, no additional tests or anonymization will be carried out on your Sample; no results will be reported and your sample, reports and data will be destroyed that have not been anonymized.

  ❑ Check here if you would like to opt out of anonymized sample retention. Note, if not checked, this is interpreted as "No"

- PerkinElmer will anonymize your data and retain the anonymized data and related anonymized reports from your Tests indefinitely for internal statistical, quality analysis, research, scientific and technical development, and market research.PerkinElmer will anonymize your data and retain the anonymized data and related anonymized reports from your Tests indefinitely for internal statistical, quality analysis, research, scientific and technical development, and market research. PerkinElmer may also share your anonymized data and anonymized report with third parties including Agios Pharmaceuticals.

  ❑ Check here if you would like to opt out of anonymized data retention. Note, if not checked, this is interpreted as "No"

For residents of NY State:
❑ By checking here I give PerkinElmer permission to store my sample for longer than 60 days. Note, if not checked, this is interpreted as "No"

RESEARCH OPTIONS

PerkinElmer may collaborate with scientists, researchers and drug developers to advance knowledge of genetic diseases. If there are opportunities to participate in future research relevant to the disease in you and/or your child, PerkinElmer may contact you or your HCP about the development of new testing, drug development, or other treatments.

WITHDRAWAL OF CONSENT

I understand this consent is voluntary and is valid until I withdraw my consent. I understand I may withdraw my consent to sample and data retention, and to the Test at any time, that PerkinElmer will not perform the Test unless I provide consent to the Test. If I withdraw any consent, it will not affect actions taken before I withdrew my consent, including any anonymization of data or of my Sample. I understand that if I wish to withdraw my consent I should contact PerkinElmer via email at: Genomics@perkinelmer.com or toll-free by telephone +1-866-354-2910 to request withdrawal.

PATIENT CONSENT TO TESTING

❑ By checking this box I attest:

I have read and understood the Informed Consent Form in its entirety, including the explanation of why my sample is being tested, how genetic testing is performed and the risks associated with genetic testing. I have had the opportunity to ask my HCP questions about the information contained herein, and understand that I am entitled to a copy of this ICF. My signature below acknowledges my free consent to the Test, and to any additional consents indicated above, and such testing in no way guarantees my health, the health of an unborn child, or the health of other family members.

Patient Signature (or Parent/Guardian if patient is minor)	Date
Patient Name	Name and Relationship (Parent/Guardian if patient is minor)

FAMILY MEMBER CONSENT TO TESTING (if applicable)

❑ By checking this box I attest: I have read and understood the Informed Consent Form in its entirety, including the explanation of why my sample is being tested, how genetic testing is performed and the risks associated with genetic testing. I have had the opportunity to ask my HCP questions about the information contained herein, and understand that I am entitled to a copy of this ICF. My signature below acknowledges my free consent to the Test, and to any additional consents indicated above, and such testing in no way guarantees my health, the health of an unborn child, or the health of other family members.

Family Member Signature	Date	Family Member Name	Relationship to Patient

FAMILY MEMBER CONSENT TO TESTING (if applicable)

❑ By checking this box I attest: I have read and understood the Informed Consent Form in its entirety, including the explanation of why my sample is being tested, how genetic testing is performed and the risks associated with genetic testing. I have had the opportunity to ask my HCP questions about the information contained herein, and understand that I am entitled to a copy of this ICF. My signature below acknowledges my free consent to the Test, and to any additional consents indicated above, and such testing in no way guarantees my health, the health of an unborn child, or the health of other family members.

Family Member Signature	Date	Family Member Name	Relationship to Patient

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