PREVENTION GENETICS
part of EXACT SCIENCES

navigateAPDS by Pharming

PREVENTIONGENETICS USE ONLY

All testing must be ordered by a qualified Healthcare Provider

THIS FORM MUST ACCOMPANY ALL SPECIMENS

FOR PATIENT SELF COLLECTING A SAMPLE, CHOOSE ONE:
☐ Ship one Saliva GeneFiX™ Saliva Collection kit to patient's address.
☐ Ship one Buccal OCD-100 kit to patient's address.

SPECIAL PROJECT - TEST REQUISITION FORM
SP312 PHARMING HEALTHCARE INC. navigateAPDS
SPONSORED TESTING PROGRAM

PERSON COMPLETING FORM	CONTACT (PHONE AND EMAIL)	DATE OF REQUEST (MM/DD/YYYY)

PATIENT INFORMATION

LAST (FAMILY) NAME	FIRST NAME	MI	DATE OF BIRTH (MM/DD/YYYY)

ADDRESS	CITY	STATE*	ZIP

EMAIL*

PHONE NUMBER*

GEOANCESTRY / ETHNICITY
☐ Asian
☐ Black/African American
☐ White/Caucasian
☐ Ashkenazi Jewish
☐ Hispanic
☐ Native American
☐ Pacific Islander
☐ French Canadian
☐ Sephardic Jewish
☐ Mediterranean
☐ Other: _____

MEDICAL RECORD NUMBER (MRN)

SPECIMEN COLLECTION DATE (MM/DD/YYYY)
If no collection date is provided, date of receipt will be used.

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

SPECIMEN SOURCE
☐ Blood ☐ Saliva
☐ Buccal ☐ Extracted DNA

BIOLOGICAL SEX
☐ Male ☐ Female
☐ Other _____ SPECIFY KARYOTYPE

HAS PATIENT'S RELATIVE BEEN TESTED AT PreventionGenetics?
☐ NO ☐ YES, provide
NAME _____ DATE OF BIRTH _____
RELATIONSHIP TO PATIENT _____ or PreventionGenetics ID NUMBER _____

BLOOD TRANSFUSION
☐ NO ☐ Within Last 30 Days,
MM/DD/YYYY _____
TYPE _____

BONE MARROW TRANSPLANT
☐ NO ☐ Yes, Date
MM/DD/YYYY _____

GENETIC COUNSELING

Telehealth genetic counseling with Genome Medical, a national telegenetics care provider, is available at no cost to patients through this sponsored testing program. Genetic counseling via telephone appointment is available for patients to provide information, education, support and address questions related to sponsored genetic testing and results.

By checking the following boxes, my patient has agreed to allow PreventionGenetics to facilitate the provision of pre-test and/or post-test genetic counseling services by Genome Medical.
- ☐ **Pre-test** genetic counseling referral to Genome Medical
- ☐ **Post-test** genetic counseling referral to Genome Medical

Genome Medical will contact the patient to schedule their genetic counseling appointment using the phone number and email address provided above.

Patients will receive a text message to schedule an appointment if they have SMS texting available on their phone.

*** Email or Phone AND State must be provided above for Genetic Counseling.**

If Power of Attorney for medical decisions/ communication is needed, the patient/family will need to provide documentation to Genome Medical upon scheduling their GC appointment.

navigateAPDS SPONSORED TESTING PROGRAM ELIGIBILITY CRITERIA

1. navigateAPDS SPONSORED TESTING PROGRAM

For individuals that meet the eligibility criteria below and wish to receive the program specific genetic testing panels.
REQUIRED: You must select below the appropriate eligibility criteria for this patient.

This program is available to patients in the U.S., Puerto Rico and Canada who meet any two or more of the following criteria below:

Clinical Features
- ☐ Bronchiectasis
- ☐ Lymphadenopathy for greater than one month
- ☐ Chronic hepatomegaly or chronic splenomegaly
- ☐ Severe, persistent, or recurrent Herpesviridae infections (e.g., EBV, cytomegalovirus)
- ☐ Enteropathy
- ☐ Lymphoma at 0-25 years - meets the 2 eligibility criteria
- ☐ Lymphoma at ≥ 26 years of age - requires second eligibility criteria

Laboratory
- ☐ Elevated levels of immunoglobulin M
- ☐ Increased number of follicular helper T cells
- ☐ Reduced number of naïve B cells

History
- ☐ Common Variable Immune Deficiency (CVID) phenotype or direct family member with CVID phenotype
- ☐ Relative with *PIK3CD* or *PIK3R1* genotype (first or second degree)

  Provide the specified details from your relative's clinical report under the requested variant section at the bottom of page 2 - meets the 2 eligibiliy criteria

REQUIRED This patient currently participates in a governmental insurance program (e.g., Medicare, Medicaid, TRICARE, etc.) ☐ Yes ☐ No
Participation in governmental insurance programs does not affect eligibility for the sponsored testing program

2. Family Targeted Variant Testing

For relatives of program participants who received a *PIK3CD* or *PIK3R1* Pathogenic, Likely Pathogenic, or Variant of Uncertain significance result who wish to receive Family Targeted Variant Testing at no additional charge. Relatives do not need to meet the eligibility criteria listed above.

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Test information is available on our website:
PreventionGenetics.com

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CLINICAL HISTORY

FAMILY HISTORY

Is there a family history of disease for which the patient is being tested? ☐ Yes ☐ No If yes, describe below and attach pedigree and/or clinical notes.

Relative's relationship to this patient	Maternal or Paternal	Diagnosed condition	Age at diagnosis

PERSONAL HISTORY

Is/was this patient affected or symptomatic?* ☐ Yes ☐ No

Provide details in the required clinical history questions (if applicable).

* Symptomatic means this patient has features or signs known or suspected to be related to the genetic testing being ordered and could include findings on physical examination, laboratory tests, or imaging.

REQUIRED CLINICAL HISTORY

Age of symptom onset: _____

Current Diagnoses: _____

Weight: _____

	Yes	No	Unknown
Familial history of APDS	☐	☐	☐
Family history of Common Variable Immune Deficiency (CVID)	☐	☐	☐

Infectious Complications

	Yes	No	Unknown
Documented severe recurrent sinopulmonary infections	☐	☐	☐
Recurrent pneumonia	☐	☐	☐
Bronchiectasis	☐	☐	☐
Severe, persistent or recurrent herpesvirus infection (e.g. EBV, CMV)	☐	☐	☐

Noninfectious Complications	Yes	No	Unknown
Lymphadenopathy for greater than one month	☐	☐	☐
Splenomegaly	☐	☐	☐
Hepatomegaly	☐	☐	☐
Nodular mucosal lymphoid hyperplasia	☐	☐	☐
Autoimmune cytopenia	☐	☐	☐
Enteropathy	☐	☐	☐
Non food related eosinophilic esophagitis	☐	☐	☐
Lymphoma	☐	☐	☐
Developmental delay	☐	☐	☐
Allergic disorders/atopy	☐	☐	☐

Lab Findings	Yes	No	Unknown
Hypogammaglobulinemia	☐	☐	☐
Elevated IgM	☐	☐	☐
Increased transitional B-cells	☐	☐	☐
Reduced naïve B cells	☐	☐	☐
Elevated T follicular helper (Tfh) cells	☐	☐	☐

ADDITIONAL CLINICAL HISTORY (OPTIONAL)

Laboratory findings **Patient Value/Reference Range**

Serum IgG: _____/_____

Serum IgM: _____/_____

TEST SELECTION

TEST CODE	TEST NAME / DESCRIPTION
☐ **13999**	**PGmax™ Inborn Errors of Immunity/Primary Immunodeficiency Panel**
☐ **19998**	**PGmax™ Primary Immunodeficiency and Malignancy Predisposition Panel**
☐ **100**	**Targeted Sanger Sequencing** of (1) *PIK3CD* or *PIK3R1* variant (pathogenic, likely pathogenic, or variant of uncertain significance (VUS)) Gene: Variant:
☐ **200**	**Targeted Sanger Sequencing** of (2) *PIK3CD* or *PIK3R1* variants (pathogenic, likely pathogenic, or variant of uncertain significance (VUS)) Gene(s): Variant(s):

SPECIAL INSTRUCTIONS

SP312

☐ **SPECIMEN COLLECTED IN NEW YORK STATE**
Include New York State Genetic Testing Healthcare Provider Statement and New York State Non-Permitted Laboratory Test Request approval letter if test is not NY state approved. For a list of NY state approved tests, see website.

COMMENTS

PROVIDER CONSENT

By signing below, you, the Healthcare Provider, agree you have obtained the patient's (or parent/guardian's if patient is a minor) informed consent to perform this test, and confirm the patient has been appropriately counseled and understands the risks, benefits, and limitations of this genetic testing and the implications of the results. You further confirm the patient authorizes PreventionGenetics to use and disclose de-identified patient test data and results ("De-identified Data") to promote research and improve the diagnosis and treatment of the genetic diseases. The De-identified Data may be used for research purposes as well as to facilitate and improve the diagnosis of genetic changes and diseases in other patients. For these reasons, PreventionGenetics may disclose De-identified Data with external physicians, scientists, researchers and pharmaceutical companies. No protected health information will be shared. As the Healthcare Provider, you hereby authorize PreventionGenetics to share your name, institution, address, and contact information with Pharming Healthcare, Inc., and consent to Pharming Healthcare, Inc. contacting you. As the Healthcare provider you attest that you will not seek reimbursement for this sponsored test from any third party, including but not limited to government healthcare programs; that participation in this sponsored testing program will not influence your medical decisions; that you are not obligated to purchase or prescribe any product or service offered by the sponsor of the Program; that you are not obligated to participate in or to encourage patients to participate in any clinical trial or other research program conducted by a sponsor; and your participation in the Program in accordance with applicable laws. For California clinicians only: I have the right to opt-out of certain uses of my data.

_____ **HEALTHCARE PROVIDER SIGNATURE**

_____ **PRINTED NAME**

_____ **DATE**

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PROVIDER INFORMATION AND REPORTING

Our preferred method of report transmission is uploading to our secure web portal, myPrevent. Please provide an email address, when possible. If you have additional specific reporting requests, indicate them BELOW.

PROVIDER INFORMATION

INSTITUTION

ADDRESS	**CITY**	**STATE**	**ZIP**

REQUESTING PHYSICIAN (First, Last, Degree)	**REQUESTING GENETIC COUNSELOR OR ALLIED PROVIDER** (First, Last, Degree)
EMAIL ADDRESS (For report access via myPrevent)	**EMAIL ADDRESS** (For report access via myPrevent)
PHONE NUMBER / **NPI# (US ONLY)**	**PHONE NUMBER** / **NPI# (US ONLY)**

IF YOU REQUIRE REPORTS TO BE TRANSMITTED VIA ANOTHER SECURE METHOD, SPECIFY HERE.

LIST ADDITIONAL HEALTHCARE PROVIDERS AND THEIR EMAILS TO ALLOW ACCESS TO REPORTS

INSTITUTIONAL BILLING

BILLING ID	**SPECIAL PROJECT NUMBER**
PHARMIN10312	**SP312**

SPECIMEN REQUIREMENTS / SHIPPING AND HANDLING INSTRUCTIONS

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. Specimen deliveries are accepted Monday-Saturday for all specimen types. Holiday schedules will be posted on our website at least one week prior to major holidays.

WHOLE BLOOD
Requirements: Collect 3 ml - 5 ml of whole blood in EDTA (purple top tube) or ACD (yellow top tube), minimum 1 ml for small infants. Heparin (green top tube) is strongly discouraged.

Shipping: At room temperature or refrigerated, a blood specimen is stable for up to 8 days. Include a refrigerated gel pack in the shipping container. Fresh blood specimens are preferred.

SALIVA
Requirements: Oragene™ or GeneFiX™ Saliva Collection kit used according to manufacturer instructions. DNA from saliva specimens is invariably contaminated with microbial and food DNA, which can impact specimen quality and may result in delayed testing and/or the need for a second specimen.

Additional instructions to help families collecting samples at home are included in each home saliva kit order.

Shipping: Specimens may be shipped at room temperature.

BUCCAL SWAB (OCD-100 Preferred)
Requirements: OCD-100 Buccal Swab used according to manufacturer instructions. Buccal swabs are most appropriate for targeted, known variant testing. DNA from buccal specimens is invariably contaminated with microbial and food DNA, which can impact specimen quality and may result in delayed testing and/or the need for a second specimen.

OCD-100 instructions are available in about 30 different languages. To request special instructions for patients, add a note in the Comments section of the kit order indicating which language is needed and we will do our best to accomodate. Default instructions are English.

Shipping: At room temperature, an OCD-100 buccal specimen is stable for up to 80 days. Specimens may be shipped at room temperature.

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

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

Comment SP312