

This requisition form can be used to submit an order for the **navigateAPDS Sponsored Testing Program**, a sponsored testing program for genetic disorders brought to you by **Pharming Healthcare, Inc.**

INSTRUCTIONS: Review the ordering options and then complete all sections of this form. Your ordering option will be indicated in the test selection section.

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. and Canada who meet ***any two or more*** of the following bulleted criteria below:

Clinical Features

- Onset of symptoms under 12 years of age
- Documented severe recurrent sinopulmonary infections (> 2 events within 3 years of each other)
- Bronchiectasis
- Lymphadenopathy for greater than one month
- Any nodular lymphoid hyperplasia
- Chronic hepatomegaly or chronic splenomegaly
- Severe, persistent, or recurrent Herpesviridae infections (e.g., EBV, cytomegalovirus)
- Autoimmune cytopenia
- Enteropathy
- Lymphoma at 0-25 years - meets the 2 eligibility criteria
- Lymphoma at ≥ 26 years of age - requires second eligibility criteria

Laboratory

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

History

- Primary Immune Deficiency diagnosis
- 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

2. GENE-SPECIFIC FAMILY FOLLOW-UP TESTING

For relatives of program participants who received a Pathogenic/Likely Pathogenic result or approved VUS who want to receive gene specific family follow-up testing at no additional charge. Relatives do not need to meet the eligibility criteria listed above. Learn more at www.invitae.com/family.

PATIENT INFORMATION

First name		MI	Last name	
Date of birth (MM/DD/YYYY)		Biological sex <input type="radio"/> M <input type="radio"/> F	MRN (medical record number)	
Ancestry <input type="radio"/> Asian <input type="radio"/> Black/African American <input type="radio"/> White/Caucasian <input type="radio"/> Ashkenazi Jewish <input type="radio"/> Hispanic <input type="radio"/> Native American <input type="radio"/> Pacific Islander <input type="radio"/> French Canadian <input type="radio"/> Sephardic Jewish <input type="radio"/> Mediterranean <input type="radio"/> Other: _____				
Phone		Email address (report access after clinician releases)		
Address		City		
State/Prov		ZIP/Postal code	Country	
Ship a kit to this patient (optional) by faxing or emailing this completed form to Invitae Kit type: <input type="radio"/> Buccal swab kit <input type="radio"/> Saliva kit Ship to: <input type="radio"/> Address above <input type="radio"/> Alternate address: _____				

SPECIMEN INFORMATION

Specimen type: Blood (3-mL purple EDTA) **-OR-** Buccal Swabs (OCD-100, 2 devices)
-OR- Saliva (Oragene™) **-OR-** DNA source: _____

We are unable to accept blood/buccal/saliva from patients with:
 • Allogeneic bone marrow transplants • Blood transfusion <2 weeks prior to specimen collection

Specimen collection date (MM/DD/YYYY): / /
If not provided, the day before specimen receipt will be used

Special cases: History of/current hematologic malignancy in patient

CLINICIAN INFORMATION

Organization name		
Phone		Fax
Address		City
State/Prov	ZIP/Postal code	Country
Primary clinical contact name (if different from ordering provider)		NPI
Primary clinical contact email address (for report access)		
Ordering provider (select <u>one</u> ordering provider by marking the checkbox before the name)		
Name	NPI	Email address (for report access)
<input type="radio"/>	_____	_____
<input type="radio"/>	_____	_____
Additional clinical or laboratory contacts (optional, to share access to order online)		
<input type="radio"/> Share this order with the primary clinical contact's default clinical team, manage at invitae.com		
Name	Email address (for report access)	
Name	Email address (for report access)	

POST-TEST GENETIC COUNSELING (please review)

Pharming sponsors post-test genetic counseling, regardless of test result type, at no additional charge to the patient through GeneMatters, an independent genetic counseling service. Please check the box below if you would like GeneMatters to contact your patient for genetic counseling once the patient's test is complete.

- I authorize Invitae to grant GeneMatters (gca@gene-matters.com) access to the patient's Invitae record for the purposes of providing genetic counseling services

INVITAE PARTNER CODE **APDS**

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	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: _____

	Y	N	UNKNOWN
Familial history of APDS	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Family history of Common Variable Immune Deficiency (CVID)	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

Infectious Complications

Documented severe recurrent sinopulmonary infections	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Recurrent pneumonia	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Bronchiectasis	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Severe, persistent or recurrent herpesvirus infection (e.g. EBV, CMV)	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

Noninfectious Complications

Lymphadenopathy for greater than one month	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Splenomegaly	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Hepatomegaly	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Nodular mucosal lymphoid hyperplasia	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Autoimmune cytopenia	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Enteropathy	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Non food related eosinophilic esophagitis	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Lymphoma	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Developmental delay	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Allergic disorders/atopy	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

Lab findings	Y	N	UNKNOWN
Hypogammaglobulinemia	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Elevated IgM	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Increased transitional B-cells	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Reduced naïve B cells	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Elevated T follicular helper (Tfh) cells	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

ADDITIONAL CLINICAL HISTORY (OPTIONAL)
Laboratory findings Patient Value/Reference Range

Serum IgG: _____/_____

Serum IgM: _____/_____

OPTIONAL - REQUESTED VARIANTS FOR THIS PATIENT'S REPORT, IF KNOWN

To have the presence or absence of specific variants commented on in this patient's report, provide the details below.

 Was the proband (individual with variant) tested at Invitae? Yes, Invitae Order ID: RQ# _____ No: Attach copy of lab results (required)

Variant(s) (e.g. GENE c.2200A>T (p.Thr734Ser) NM_00012345) If left blank, all variants identified in the proband will be commented on.

This patient's relationship to proband:
 Parent Sibling Grandchild

 Child Self Other: _____

