PREVENTION GENETICS part of EXACT SCIENCES

Test information is available on our website:
PreventionGenetics.com

PREVENTIONGENETICS USE ONLY

All testing must be ordered by a qualified Healthcare Provider

THIS FORM MUST ACCOMPANY ALL SPECIMENS

SPECIAL PROJECT - TEST REQUISITION FORM

SP144 - CHOLESTASIS 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)

PATIENT ID

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

GEOANCESTRY / ETHNICITY
- ☐ White/Caucasian
- ☐ Hispanic
- ☐ Black/African American
- ☐ Native American/First Nations
- ☐ East Asian
- ☐ South Asian
- ☐ Middle Eastern
- ☐ Ashkenazi Jewish
- ☐ Other: _____
- ☐ Not Specified

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

SPECIMEN SOURCE
- ☐ Blood
- ☐ Buccal

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,

DATE (MM/DD/YYYY) _____

TYPE _____

BONE MARROW TRANSPLANT
- ☐ NO ☐ Yes, include date

DATE (MM/DD/YYYY) _____

RELEVANT CLINICAL INFORMATION. We strongly encourage the inclusion of detailed clinical notes/completion of the clinical data checklist and a pedigree. The ability to interpret variants directly correlates with the quality of clinical information provided.

☐ Clinical records attached.

ADDITIONAL RELEVANT CLINICAL INFORMATION:

Does patient have chronic cholestasis (evidence of laboratory test abnormalities >3 months)
- ☐ No
- ☐ Yes. Date abnormal liver tests first identified (MM/DD/YYYY) _____

Which of the following liver test abnormalities does the patient currently have (CHECK ALL THAT APPLY)
- ☐ ALT
- ☐ GGT
- ☐ AST
- ☐ Direct/conjugated bilirubin
- ☐ Serum bile acids
- ☐ Alkaline phosphatase
- ☐ No liver test abnormalities

Does the patient have any of the following complications of chronic liver disease? (CHECK ALL THAT APPLY)
- ☐ Signs/symptoms of cirrhosis or portal hypertension
- ☐ Growth failure
- ☐ Fat-soluble vitamin deficiency
- ☐ Impaired liver function (e.g., INR not corrected by vitamin K, low albumin)
- ☐ Pruritus
- ☐ Liver cancer or cholangiocarcinoma
- ☐ None

Family history of liver disease
- ☐ No
- ☐ Yes, briefly describe _____

Does the patient have any extra-hepatic manifestations
- ☐ No
- ☐ Yes, briefly describe _____

PATIENT ELIGIBILITY AND TEST SELECTION

CRITERIA FOR FREE TESTING:

Patient must meet one of the criteria below:
- ☐ Patient is currently cholestatic, or has a history of cholestasis, without an identified cause
- ☐ Unexplained chronic liver disease
- ☐ Unexplained chronic diarrhea plus tendon xanthomas, neurological deterioration, or idiopathic cataracts
- ☐ First degree relative with cholestasis and confirmed variant/s in the following diseases/genes:
  - Alagille syndrome: *JAG1, NOTCH2*
  - PFIC: *ABCB11, ABCB4, ATP8B1, KIF12, MYO5B, NR1H4, PLEC, PSKH1, SLC51A, TJP2, SEMA7A, USP53, VPS33B, WDR83OS, ZFYVE19*
  - CTX: *CYP27A1*

AND Patient must meet all criteria below:
- ☐ Extrahepatic disorders are not a consideration (eg, biliary atresia, choledochal cyst, large duct PSC)
- ☐ TPN cholestasis is not suspected as a primary diagnosis
- ☐ Patient lives in the U.S. or Canada.

TEST CODE	DESCRIPTION	ADDITIONAL INFORMATION
☐ 16229	**Cholestasis Sequencing Panel**	NextGen sequencing and CNV detection (see Sponsored Testing page or program brochure for gene list)

COMMENTS

SPECIAL INSTRUCTIONS

SP144

☐ **SPECIMEN COLLECTED IN NEW YORK STATE**

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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.

Provide the patient's phone number and email address to enable Genome Medical to contact the patient to schedule their genetic counselor appointment.

PATIENT PHONE NUMBER

PATIENT EMAIL ADDRESS

U.S. STATE WHERE PATIENT RESIDES (REQUIRED)

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

- If Power of Attorney for medical decisions/ communication is needed, please provide acopy to Genome Medical at clinical@genomemedical.com.
- For other questions related to counseling, Genome Medical can be reached at: clinical@genomemedical.com.

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 anonymize and share test data and results to promote research and improve the diagnosis and treatment of genetic diseases. The data and results 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 anonymize and share test data and results with external physicians, scientists, researchers and pharmaceutical companies. No personal identifying information will be shared.

As the Healthcare Provider, you hereby authorize PreventionGenetics to share your name, institution, address, and contact information with the pharmaceutical company program sponsors, and consent to program sponsors contacting you.

FOR CALIFORNIA DOCTORS ONLY: To opt-out of the sharing of provider contact information with other healthcare entities, check this box ☐

HEALTHCARE PROVIDER SIGNATURE | PRINTED NAME | DATE

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.

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 **MIRUMP10144**

SPECIAL PROJECT NUMBER **SP144**

SPECIMEN REQUIREMENTS - SHIPPING AND HANDLING INSTRUCTIONS

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.

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. If frozen, a blood specimen is stable for up to 1 month before shipping. Frozen blood specimens should be shipped frozen (preferably on dry ice) overnight.

☐

☐ mirum

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 accommodate. 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.

CONTACT US

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.

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

REFERENCE SP144

This program may be canceled or changed at any time.