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

SP102 - PEROXISOMAL BIOGENESIS DISORDER - ZELLWEGER SPECTRUM DISORDER (PBD-ZSD)

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

PATIENT INFORMATION

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

PATIENT ID	BIOLOGICAL SEX	BLOOD TRANSFUSION	BONE MARROW TRANSPLANT
	☐ Male ☐ Female ☐ Other	☐ NO ☐ Within last 30 days, include:	☐ NO ☐ Yes, include date
GEOANCESTRY / ETHNICITY	SPECIFY KARYOTYPE	DATE (MM/DD/YYYY)	DATE (MM/DD/YYYY)
SPECIMEN COLLECTION DATE (MM/DD/YYYY) If no collection date is provided, date of receipt will be used.	**SPECIMEN SOURCE** ☐ Whole Blood	TYPE	

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.

TEST SELECTION

TEST CODE	DESCRIPTION	ADDITIONAL INFORMATION	SPECIAL INSTRUCTIONS
12655	**PBD-ZSD Sequencing Panel**	NextGen sequencing with Copy Number Variant (CNV) detection is performed for the following 13 genes: *PEX1, PEX2, PEX3, PEX5, PEX6, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19,* and *PEX26.*	**SP102** ☐ **SPECIMEN COLLECTED IN NEW YORK STATE**
Auto-Reflex If 12655 is positive or suspicious	**DHCA/THCA Biochemical Testing**	If the PBD-ZSD panel is positive or suspicious, reflex formed at Cincinnati biochemical testing will be per Children's Hospital Medical Center (CCHMC) to help identify the presence and concentration levels of DHCA/THCA, atypical bile acids which can be hepatotoxic and associated with liver disease in PBD-ZSD.	

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.

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PROGRAM ELIGIBILITY AND PROVIDER AUTHORIZATION

Patients must meet one of the criteria below:

- ☐ Diagnosed with peroxisomal biogenesis disorder-Zellweger spectrum disorder (PBD-ZSD)
- ☐ Clinical suspicion of peroxisomal biogenesis disorder-Zellweger spectrum disorder (PBD-ZSD) (e.g. neurological, vision, hearing, or hepatic deterioration)

By signing below, you, the Healthcare Provider, agree you have obtained the patient's 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 Mirum Pharmaceuticals, and consent to Mirum Pharmaceuticals 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 CONTACT 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#	PHONE NUMBER	NPI#

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
MIRUMPH10102	**SP102**

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.

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 SP102

mirum

This program may be canceled or changed at any time.