

SPECIAL PROJECT - TEST REQUISITION FORM

SP101 - X-LINKED ADRENOLEUKODYSTROPHY (X-ALD)

NEWBORN SCREENING REFLEX TEST PROGRAM

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

LAST (FAMILY) NAME	FIRST NAME	MI	DATE OF BIRTH (MM/DD/YYYY)
PATIENT ID	BIOLOGICAL SEX <input type="checkbox"/> Male <input type="checkbox"/> Female <input type="checkbox"/> Other	BLOOD TRANSFUSION <input type="checkbox"/> NO <input type="checkbox"/> Within last 30 days, include: DATE (MM/DD/YYYY)	BONE MARROW TRANSPLANT <input type="checkbox"/> NO <input type="checkbox"/> Yes, include date DATE (MM/DD/YYYY)
GEOANCESTRY / ETHNICITY	SPECIFY KARYOTYPE		
SPECIMEN COLLECTION DATE (MM/DD/YYYY) If no collection date is provided, date of receipt will be used.	SPECIMEN SOURCE <input type="checkbox"/> Whole Blood <input type="checkbox"/> Buccal	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
12671	X-ALD Newborn Screening Reflex Panel	NextGen sequencing with Copy Number Variant (CNV) detection is performed for the following 26 genes: <i>ABCD1, ACBD5, ACOX1, ADAR, BCAP31, HSD17B4, IFIH1, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, TREX1</i>	SP101 <input type="checkbox"/> SPECIMEN COLLECTED IN NEW YORK STATE

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 messaging 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.

PROGRAM ELIGIBILITY AND PROVIDER AUTHORIZATION

Patients must meet ONE of the criteria below:

- ☐ Testing will be offered to children who screened positive for X-ALD on initial state newborn screening.
- ☐ Testing will be offered to a blood relative (sibling, cousin) of a PBD-ZSD positive patient who was tested at PreventionGenetics.

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

MIRUMPH10101

SPECIAL PROJECT
NUMBER

SP101

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.

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 SP101



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This program may be canceled or changed at any time.