



# UNCOVERING RARE OBESITY®

Welcome to **Uncovering Rare Obesity**, a no-charge\* genetic testing program for rare genetic diseases of obesity

## Step-by-Step Testing Guide

This guide will help you to start testing your patients for rare genetic diseases of obesity through **Uncovering Rare Obesity**.

Educational and support materials for you and your patients are available at [UncoveringRareObesity.com](http://UncoveringRareObesity.com).

### The program has 3 main steps:

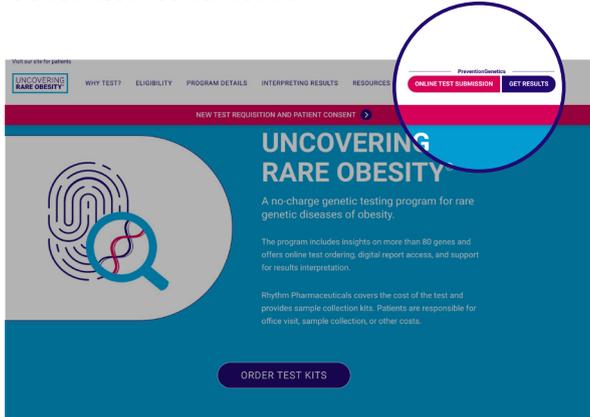
- 1 **CREATE**  
Create your account and login
- 2 **TEST (IN-OFFICE AND AT-HOME OPTIONS)**  
Order kits and submit tests
- 3 **TRACK**  
Track test status and view results

\*Eligible individuals can receive a genetic test and 2 genetic counseling sessions (1 before and 1 after the test) at no charge. Participants are responsible for any doctor visit, sample collection, or other costs.

- CREATE ACCOUNT
- TEST IN-OFFICE: ORDER KITS
- TEST IN-OFFICE: DIGITAL FORMS
- TEST IN-OFFICE: PAPER FORMS
- TEST AT-HOME: DIGITAL AND PAPER
- TRACK STATUS & VIEW REPORTS

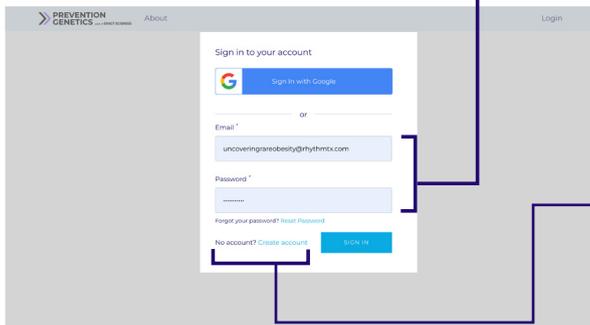
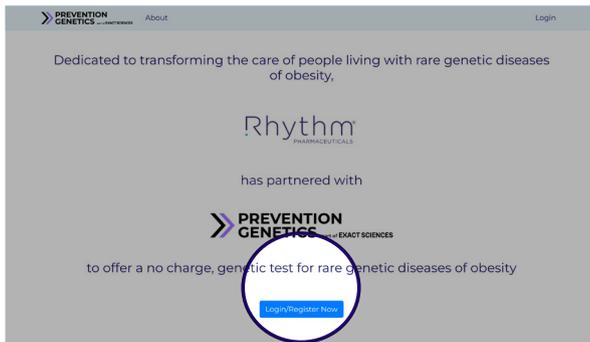
# Accessing the Uncovering Rare Obesity Portal

## Visit the website:



- Visit [UncoveringRareObesity.com](https://UncoveringRareObesity.com)
  - Bookmark this page for ordering kits, submitting tests, tracking status, and accessing results
- Click **either ONLINE TEST SUBMISSION or GET RESULTS under PreventionGenetics** on the upper right to access the portal for Uncovering Rare Obesity

## Log in:

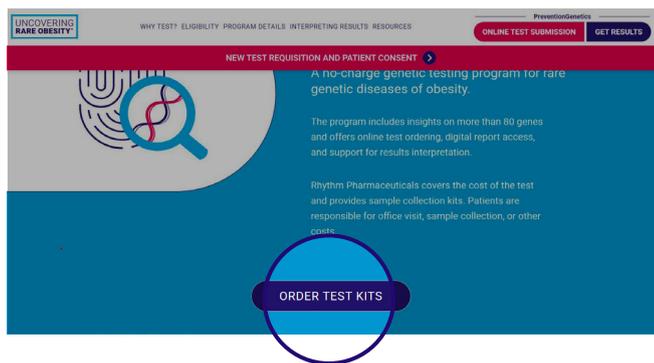


- Click **Login/Register Now**
- **Note:** Each user, including staff, should have their own account. Please do not share accounts.
- **Already a myPrevent user with PreventionGenetics?** Use your existing username and password from myPrevent with no additional sign up needed
- **Not yet a myPrevent user?** Select "Create account"
  - Please do not sign in with Google
  - Use an email address affiliated with your institution (this email address is also your username) and please confirm correct spelling of your email
  - Use the same email address on the Test Requisition Form
  - Staff members assisting with the program should create their own accounts
  - You will receive an email to activate your account

## Order Office Kits

Follow the directions below for sending test kits to your office. Do not send these test kits home with patients. **For at-home testing, go to page 9.**

### Order office kits:



- Click on **ORDER TEST KITS** at [UncoveringRareObesity.com](https://www.uncoveringrareobesity.com)

 A screenshot of the 'Office Kit Ordering' form on the PreventionGenetics website. The form is titled 'Office Kit Ordering' and includes several sections:
 

- Kit Selection:** Two dropdown menus for 'Blood' and 'Buccal'.
- Shipping and Contact Information:** Fields for First Name, Last Name, NPI Number, Email, Specialty, Designation, Practice Phone, Practice Name, Street Address, Street Address 2, City, Country (radio buttons for US, US Territories, Canada), State, and Zip Code.
- Preferred Return Shipping Method:** A dropdown menu.
- Comments:** A text area with the prompt 'If you have any comments enter them here'.
- Agreement:** A checkbox and text stating: 'By submitting this form, I certify that I am a healthcare professional licensed in the United States or its territories, or Canada, and I agree to the Terms of Use of this website. I understand that this information will be shared with Rhythm. For information regarding Rhythm's privacy practices, please refer to Rhythm's Privacy Policy.'
- Submit:** A blue button at the bottom left.

 The footer contains contact information for PreventionGenetics and the Rhythm logo.

- Click on **Order Office Kits**
- Enter quantity of “Blood” or “Buccal” under “Kit Selection”
- Select “Preferred Return Shipping Method” (FedEx or UPS)
- Fill out the required fields and click **Submit**

**Once you SUBMIT your order, kits will arrive in 3-5 business days.**

## Submit Test: Digitally

Digital test submissions include the Test Requisition and Patient Consent/Authorization Forms. At the end of the test submission, these forms must be printed, signed, dated, and returned in the envelope with the sample.

### Enter patient information:

The screenshot shows the 'RARE GENETIC OBESITY PANEL - TEST REQUISITION FORM' on the Prevention Genetics website. The 'Submit Test' button is circled in blue. The form includes a progress bar, a 'Next' button, and radio buttons for sample collection methods: 'In office sample collection', 'At home sample collection', and 'Repeat testing using DNA at lab'.

- To ensure you are ordering the Uncovering Rare Obesity-sponsored test, double check that you are on the [Uncovering Rare Obesity portal](https://rhythm.preventiongenetics.com) at [rhythm.preventiongenetics.com](https://rhythm.preventiongenetics.com)
- Click on **Submit Test**
- Select test type
  - **In-office** sample collection
- Enter the **Patient Information** and click **Next**

## Determine eligibility:

For new patients, please select one of the following eligibility parameters below:

Patient Age: 23

Age of  $\leq 18$  years of age, BMI  $\geq 97$ th percentile.

Age of  $\geq 19$  years of age, BMI  $>$  or equal to 40, and has a history of childhood obesity.

Clinical or suspected diagnosis of Bardet-Biedl Syndrome (BBS).

Exception Requested.

In rare situations, patients present with compelling reasons for testing but do not meet above criteria. All exception requests are subject to sponsor approval. PreventionGenetics will notify you if the exception is not approved.

For family members of selected previously tested patients

Family member to previously tested proband

Test eligibility for first degree relatives will be indicated in the notes section of the proband report. If eligible, a full gene panel analysis will be performed.  
Call PreventionGenetics at (844) 513-3994 to confirm eligibility prior to test submission.

- For a new patient, select an eligibility parameter or request an exception
- If the patient qualifies for family testing, please select “Family member to previously tested proband”
  - You will need the previously tested patient’s name and PG ID found on the original report
- Click **Next**

## Enter clinical information:

**REQUIRED CLINICAL INFORMATION**

Waist Circumference  In  cm  Not Evaluated

History of weight loss interventions (check all that apply):

Diet/lifestyle

Anti-obesity medications

Bariatric Surgery

None

Unknown

Additional Comments

Family History of obesity (check all that apply):

Mother

Father

Sibling(s)

None

Unknown

Family history of genetic disease and/or earlier testing:

Yes

No

Unknown

Diagnosis of Bardet Biedl Syndrome (BBS)

Clinically Diagnosed

Suspected

Not suspected

Unknown

**ADDITIONAL CLINICAL INFORMATION**

Developmental Delay  Yes  No  Not Evaluated

Cognitive Impairment  Yes  No  Not Evaluated

• Learning Difficulties  Yes  No  Not Evaluated

• Speech Delay  Yes  No  Not Evaluated

Hypogonadism or genitourinary anomalies  Yes  No  Not Evaluated

Other features

Hyperphagia  Yes  No  Unknown

*Hyperphagia is characterized by pathological, insatiable hunger accompanied by abnormal food seeking behaviors and extreme pre-occupation with food that results in a significant negative impact on the lives and functioning of patients and caregivers.*

*Examples of behaviors associated with hyperphagia include night awakening to eat, sneaking or hiding food, being distressed if denied food, and hyperphagia may impact focus and performance at school and work.*

*Please note that behaviors may vary between patients, and not all behaviors are expected to be present in every patient.*

On a scale of 1-5, how would you categorize the severity of the hyperphagia based on the above definition?

1 (mild)

2

3

4

5 (severe)

N/A

Age of hyperphagia onset in years

0-2 years

3-5 years

6-10 years

11-17 years

Adult

Unknown

Vision Impairment  Yes  No  Not Evaluated

• Retinal Dystrophy  Yes  No  Not Evaluated

• Night blindness  Yes  No  Not Evaluated

Renal anomalies  Yes  No  Not Evaluated

History of polydactyly  Yes  No  Not Evaluated

- Enter **Required Clinical Information** and **Additional Clinical Information**
- Click **Next**

## Enter provider information:

The screenshot shows a web form titled "RARE GENETIC OBESITY PANEL - TEST REQUISITION FORM". The "PROVIDER INFORMATION" section is active. It includes a progress bar at the top with "Next" and "Back" buttons. Below the title, there are instructions: "Our preferred method of report transmission is uploading to our secure web portal. Please provide an email address, when possible. If you have additional specific reporting requests, indicate them below." The form is divided into two main sections: "PROVIDER INFORMATION" and "REPORTING INSTRUCTIONS (OPTIONAL)".

**PROVIDER INFORMATION:**

- I am...
  - The licensed healthcare provider responsible for this patient's test order
  - An allied provider or support staff entering the order on behalf of the ordering healthcare provider
- Institution: Rhythm
- Street Address: 222 Berkeley Street 12th Floor
- City: Boston
- Country:  US,  US Territories,  Canada
- State: MA
- Zip Code: 02116
- Requesting Physician or Provider:
  - Rhythm: [input]
  - Tester: [input]
  - DO: [input]
- Phone Number: 857-264-4280
- NPI#: 123456789
- Ordering Provider Email: uncoveringrareobesity@rhythmtx.com
- Specialty: Pediatric Endocrinology
- Designation: Medical Doctor

**REPORTING INSTRUCTIONS (OPTIONAL):**

- Report will be sent to: uncoveringrareobesity@rhythmtx.com
- If you require reports to be transmitted another way, specify instructions here: [text area]
- List Email Addresses of Other Providers That Should Have Access to Results. Account Required for Online Report Access.
 

Email	Email	Email
[input]	[input]	[input]

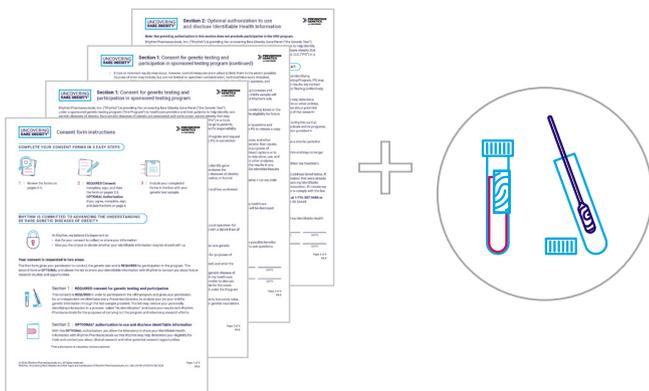
- Select provider type:
  - If you are the ordering provider, select "The licensed healthcare provider"
  - If you are ordering on behalf of a provider, select "An allied provider or support staff"
  - Enter ordering provider's email address (which is also the provider's username)
- Click **Next**

## Reporting instructions and sharing reports with other providers:

- Verify or update provider information (autofilled from your account)
- To receive alerts via a method other than email, enter the necessary information
  - All alerts come from [reports@preventiongenetics.com](mailto:reports@preventiongenetics.com)
- To share this report with other clinicians, enter their email addresses/usernames (double check spelling)
  - When a report is ready, alerts are sent to all clinicians listed
  - Email alerts include a link to the reports page on the [Uncovering Rare Obesity portal](#)
- Click **Print Form**

## Submitting forms and sample:

- Review and print
- Check the acknowledgment box and click **Submit Form**
- View confirmation of order and next steps

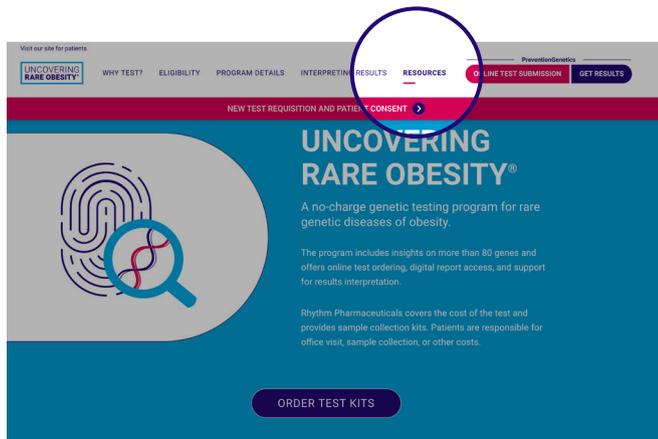


- Sign the Provider Authorization Form
- Request your patient to sign the Patient Consent Form (required) and Authorization Form (optional)
- Include signed forms and the sample and return to **PreventionGenetics** using the enclosed shipping supplies

## Submit Test: Paper

- Paper submissions include a Test Requisition Form and Patient Consent/Authorization Forms downloaded and printed from [UncoveringRareObesity.com/#resources](https://UncoveringRareObesity.com/#resources)
- Forms can be downloaded, partially prefilled, saved, and printed ahead of patient visits
- The test kit box does not include these forms

### Printing forms:



- Navigate to [UncoveringRareObesity.com/#resources](https://UncoveringRareObesity.com/#resources) to download the Test Requisition Form and the Patient Consent and Authorization Forms; click **Print**
- Ordering provider should sign and complete the Test Requisition Form and sign the Provider Authorization section
- Request your patient to sign the Patient Consent Form (required) and Authorization Form (optional)
- Please include all forms in the envelope with the sample

## Test At-Home

Follow these instructions to send a Buccal test kit to the patient's home for at-home sample collection using digital or paper forms.

### For At-Home Testing—Digital:

- Click on **Submit Test**
- Select test type
  - Select “At-home sample collection”
- Enter requested information
- Click **Submit**

- View confirmation of order submission and next steps
- Provider Authorization will be sent via DocuSign email within 1-2 business days
- Patient Consent and Authorization Forms will be sent directly to the patient in the test box with instructions for returning to the lab

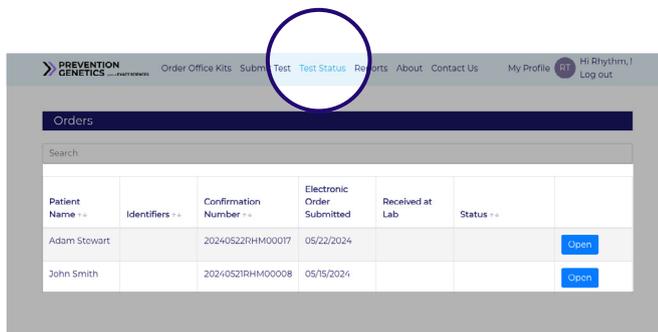
**After the ordering provider signs the DocuSign, the At-Home Test Kit will arrive at your patient's home in 3-5 business days.**

Consider following up with your patient to ensure they have received their test kit and have followed the directions to submit their test.



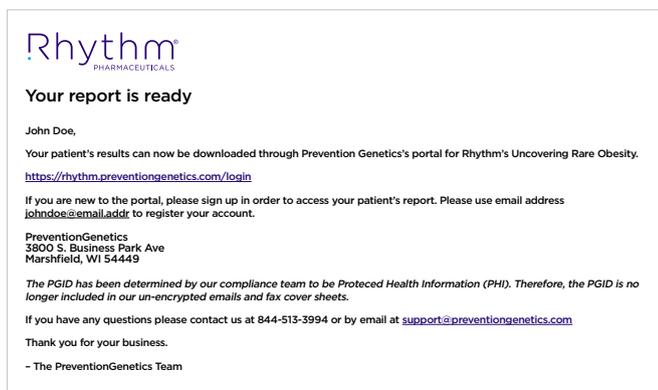
## Track Tests and View Reports

### Track the status of your submitted tests:



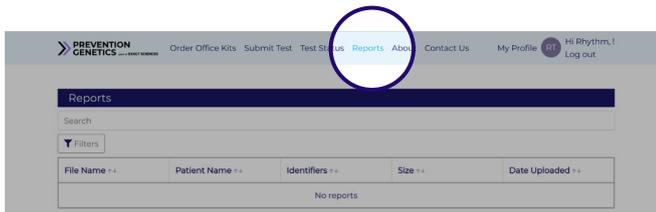
- Click on **Test Status**
- **Note:** “Awaiting Specimen” only applies to tests submitted digitally, for both in-office and at-home sample collection
- **Test status options include:**
  - Awaiting Specimen
  - Specimen Received
  - Testing In Progress
  - Testing On Hold
  - Cancelled
  - Report Ready

### Check your email for report notification:



- Results are available approximately 3 weeks after the lab receives the sample and completed forms
- When the results are ready, an email alert from [reports@preventiongenetics.com](mailto:reports@preventiongenetics.com) with a link to the account will be sent
- Add [reports@preventiongenetics.com](mailto:reports@preventiongenetics.com) to your contacts to prevent these emails from going to spam
- You may also visit [UncoveringRareObesity.com](https://UncoveringRareObesity.com) and click on **Get Results** to access patient reports through the [Uncovering Rare Obesity portal](https://UncoveringRareObesity.com) at [rhythm.preventiongenetics.com](https://rhythm.preventiongenetics.com)

## View reports:



- The **Reports** page includes a list of all tested patients
- Filter the results to narrow down the list of patients
- For more detailed instructions on accessing results, check out the [Accessing Results Guide](#) located at [UncoveringRareObesity.com/#resources](https://UncoveringRareObesity.com/#resources)

## Download the report:

Reports				
Search				
Filters				
(-)	File Name	Patient Name	Identifiers	Date Uploaded
(-)	<a href="#">Download Report</a>	John Doe	PG ID: 2023-083-076 Reference Code: 9101B674271400DAAC	05/04/2023
Reported Variants				
Gene	DNA Variations	Predicted Effects	Zygosity	Interpretation
WDPCP	c.21581G>A	GT Donor	HET	Suspected Pathogenic

- Click on **“Download Report”** to access a PDF of the test results
- To consult with a geneticist about the results of the report, call **PreventionGenetics** at **1-844-513-3994**
- Patients interested in genetic counseling may contact **Metis Genetics** at [support@metisgenetics.com](mailto:support@metisgenetics.com) or **1-844-463-8474**

## For help with the Uncovering Rare Obesity portal or questions about results

PreventionGenetics

 [support@preventiongenetics.com](mailto:support@preventiongenetics.com)

 Call: 1-844-513-3994

Fax: 715-406-4175

## For pre- and post-test genetic counseling, your patient can contact Metis Genetics

 [support@metisgenetics.com](mailto:support@metisgenetics.com)

 Call: 1-844-463-8474

## For general questions about the program or to be connected to a Rhythm representative

 [UncoveringRareObesity@rhythmtx.com](mailto:UncoveringRareObesity@rhythmtx.com)

## For program resources

[UncoveringRareObesity.com/#resources](https://UncoveringRareObesity.com/#resources)

[UncoveringRareObesity.com/patient/#resources](https://UncoveringRareObesity.com/patient/#resources)

**Sponsored by Rhythm Pharmaceuticals. DNA testing is conducted by PreventionGenetics, a CLIA-accredited clinical laboratory.**