Uncovering Rare Obesity® Genetic Testing Program: Overview and Health Care Provider Utilization

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Learn more about the expanded Uncovering Rare Obesity® program at UncoveringRareObesity.com

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Summary

- **The Uncovering Rare Obesity® testing program provides no-charge genetic testing for eligible patients in the United States who have hallmark symptoms of rare genetic diseases of obesity**
- **Since its launch, the Uncovering Rare Obesity® program has been highly utilized by health care providers across specialties for identifying and diagnosing rare genetic diseases of obesity, with 1,425 health care providers submitting 7,674 tests over the course of 22 months**

Introduction

- Some forms of obesity are caused by rare variants in one of multiple genes involved in energy and hunger regulation, such as genes in the melanocortin-4 receptor (MC4R) pathway[1-3]
  - Impaired MC4R signaling due to rare variants has been associated with early-onset, severe obesity and insatiable hunger, or hyperphagia[1,2]
- Genetic testing for obesity is recommended in patients with early-onset (ie, before 5 years of age), severe obesity and clinical features of genetic obesity, such as hyperphagia, or a family history of severe obesity[4]
- Routine genetic testing is needed to identify and diagnose patients with various rare genetic diseases of obesity[5]
  - Testing for genetic obesity may inform specialized management strategies or eligibility for clinical trials[1,4,6,7]
- The Uncovering Rare Obesity® testing program aims to enhance genetic testing access for patients with suspected rare genetic diseases of obesity in the United States[8]

Objective

- To analyze the utilization patterns of the Uncovering Rare Obesity® program since its launch

Methods

Uncovering Rare Obesity® Program

- The testing program was launched in May 2019, and the sequencing panel included 40 genes associated with obesity
  - In July 2021, the panel was expanded to include 79 genes and 1 chromosomal region; the current analysis includes data from the initial 40-gene panel
- If eligible, individuals will receive a no-charge genetic test and up to 2 genetic counseling sessions
- Individuals may be eligible for testing if they are located in the United States and its territories, and
  - Are ≤18 years of age with body mass index (BMI) ≥97th percentile, or
  - Are ≥19 years of age with BMI ≥40 kg/m^2 and have a history of childhood obesity, or
  - Had a select family member who was previously tested
    - In July 2021, the eligibility criteria were expanded to include patients showing clinical symptoms of Bardet-Biedl syndrome
- In the current analysis, blood, buccal, or saliva kits were available for sample collection
- Test kits can be requested only by licensed health care providers (HCPs)
- The test is run by a Clinical Laboratory Improvement Amendments–accredited clinical laboratory, and results are available ~3 weeks following sample receipt

Program Utilization Analysis

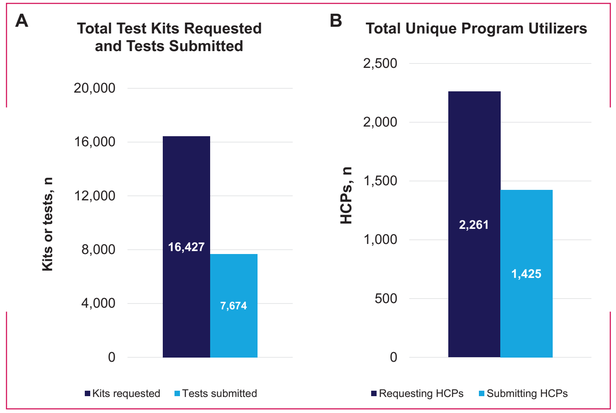
- The number of Uncovering Rare Obesity® test kit requests and submissions by HCPs were evaluated to determine trends in program utilization
- The specialties of HCPs utilizing the Uncovering Rare Obesity® program and age of patients tested were assessed

Results

Uncovering Rare Obesity® Program Utilization

- As of March 31, 2021, the Uncovering Rare Obesity® program was utilized by HCPs in all 50 states, Puerto Rico, and the District of Columbia
- In total, 16,427 test kits were requested, of which 7,674 (46.7%) had been returned for processing (Figure 1A)
  - 84.5% of tests were processed within 3 weeks of receipt by the testing laboratory; 92.5% were processed within 4 weeks
- Of 2,261 unique HCPs requesting test kits, 1,425 had submitted tests for processing, yielding a utilization rate of 63.0% (Figure 1B)
  - The mean number of test submissions per HCP was 5.4
  - The top-10 program utilizers submitted a combined average of 53 tests per month

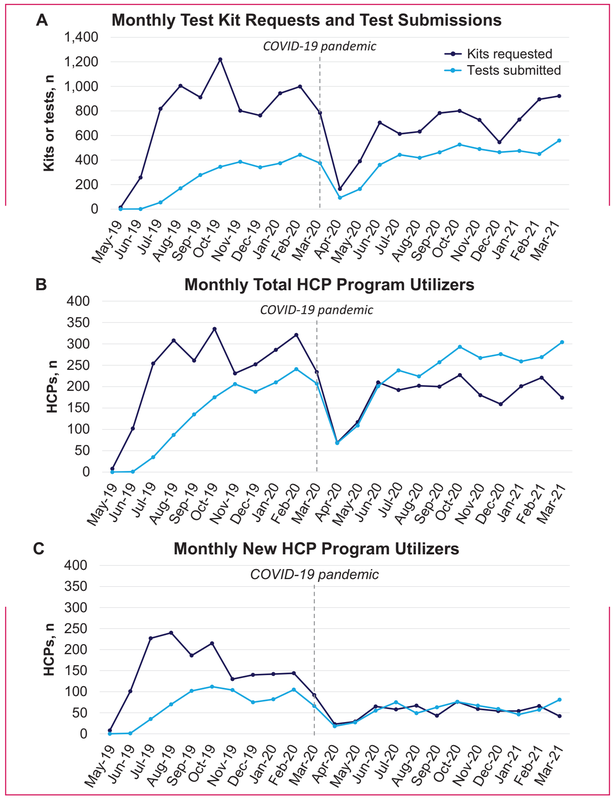
Figure 1. (**A**) Number of total Uncovering Rare Obesity® kits requested and tests submitted and (**B**) number of unique HCPs who requested kits and submitted tests from May 2019 to March 2021 excluding cancelled tests.



HCP, health care provider.

- Overall trends in Uncovering Rare Obesity® program utilization decreased in April 2020, coinciding with timing of the initial broad effects of the COVID-19 pandemic in the United States; test request, submission, and utilization rates generally increased toward prepandemic levels after June 2020 (Figure 2)
- Between October 2020 and March 2021, the monthly average of new HCP Uncovering Rare Obesity® unique program utilizers was 64

Figure 2. (**A**) Number of test kit requests and test submissions, (**B**) number of total HCP program utilizers, and (**C**) number of new HCP program utilizers by month excluding cancelled tests.



Dashed line indicates timing of the initial broad effects of the COVID-19 pandemic in the United States. HCP, health care provider.

HCP and Patient Characteristics

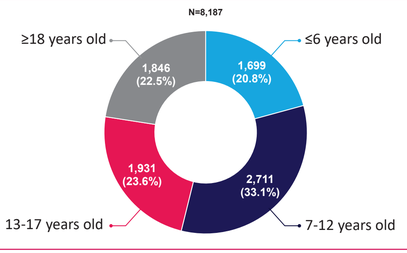
- The most common specialties of HCPs submitting tests were pediatric endocrinology, general pediatrics, and medical genetics (Table)
- The majority of tests were submitted for pediatric and adolescent patients, although many adult patients also received testing (Figure 3)

Table. Specialties of Program Utilizers (Excluding Cancelled Tests)

Specialty	HCPs, n (%) (n=1,425)	Test submissions, n (%) (n=7,674)	Average test submissions per HCP
Pediatric endocrinology	480 (33.7)	3,132 (40.8)	6.5
General pediatrics	321 (22.5)	1,717 (22.4)	5.3
Medical genetics	204 (14.3)	783 (10.2)	3.8
Endocrinology	98 (6.9)	461 (6.0)	4.7
Family practice	78 (5.5)	333 (4.3)	4.3
All other specialties	245 (17.2)	1,249 (16.3)	5.1

HCP, health care provider.

Figure 3. Age distribution of patients with tests submitted to the Uncovering Rare Obesity® program between May 2019 and March 2021.



Conclusions

- The Uncovering Rare Obesity® program has been highly utilized by HCPs in the United States since its launch
- The program may serve as a useful tool to enhance genetic testing access and improve the diagnosis of patients with some rare genetic diseases of obesity, which may inform specialized management strategies or clinical trial eligibility

*Alastair Garfield was an employee of Rhythm Pharmaceuticals, Inc., at the time of abstract submission.

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Acknowledgments: This study was sponsored by Rhythm Pharmaceuticals, Inc. Assistance with preparation of this poster was provided under the direction of the authors by Rhyomi Sellnow, PhD, and David Boffa, ELS, of MedThink SciCom and funded by Rhythm Pharmaceuticals, Inc.

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