Are you aware of the frequency of obesity-associated gene variants?

It is likely that the true prevalence of rare genetic diseases of obesity has previously been underestimated because genetic testing is often not done in individuals with obesity

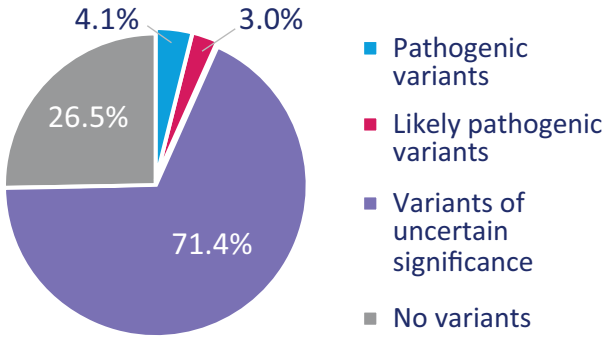
Uncovering Rare Obesity® is a no-charge*, extensive genetic testing program for rare genetic diseases of obesity sponsored by Rhythm Pharmaceuticals, Inc.

To be eligible for testing through the Uncovering Rare Obesity® program, patients must be located in the United States or its territories, or Canada, and:

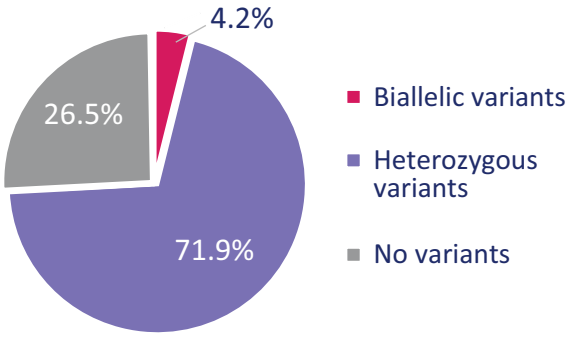
- ≤18 years of age with body mass index (BMI) ≥97th percentile, OR
- ≥19 years of age with BMI ≥40 kg/m^2 and a history of childhood obesity, OR
- an immediate family member of select, previously tested patients, OR
- showing clinical symptoms of Bardet-Biedl syndrome (BBS)

*Rhythm Pharmaceuticals covers the cost of the test and provides sample collection kits. Patients are responsible for any office visit, sample collection, or other costs.

Sequences from 11,671 individuals were analyzed using the expanded 79-gene and 1–chromosomal region panel[a]



73.5% of individuals had at least 1 pathogenic variant, likely pathogenic variant, or variant of uncertain significance in one of the 79 genes.



73.5% of individuals had biallelic or heterozygous variants. Includes homozygous and presumed compound heterozygous variants.

Of the total population, 2.9% of individuals carried pathogenic or likely pathogenic variants that also met the mode of inheritance[b]

[a]Patients may have >1 variant and therefore may be represented in >1 section of the pie charts. [b]Mode of inheritance criteria were defined as ≥2 alleles in autosomal recessive conditions or ≥1 allele in autosomal dominant conditions.

Most frequent variants:

ALMS1 - 9.22% (n=1076)
CEP290 (*BBS14*) - 4.87% (n=568)
PLXNA4 - 3.50% (n=408)
MC4R - 3.49% (n=407)
PLXNA1 - 3.44% (n=401)
IFT172 (*BBS20*) - 3.41% (n=398)
RPGRIP1L - 3.34% (n=390)
PLXNA3 - 3.18% (n=371)
PLXNA2 - 3.10% (n=362)
SH2B1 - 2.58% (n=301)
LEPR - 2.51% (n=293)
MAGEL2 - 2.49% (n=291)
MKS1 (*BBS13*) - 2.41% (n=281)
CREBBP - 2.26% (n=264)
SEMA3G - 2.23% (n=260)
POMC - 2.13% (n=249)
BBS9 - 2.02% (n=236)
NCOA1 - 2.02% (n=236)
BBS1 - 1.98% (n=231)
PCSK1 - 1.92% (n=224)

Uncovering Rare Obesity® panel

ADCY3
AFF4
ALMS1
ARL6 (*BBS3*)
BBIP1 (*BBS18*)
BBS10
BBS12
BBS1
BBS2
BBS4
BBS5
BBS7
BBS9 (*PTHB1*)
BDNF
CFAP418 (*BBS21*)
CEP290 (*BBS14*)
CPE
CREBBP
CUL4B
DNMT3A
DYRK1B
EP300
GNAS
HTR2C
IFT172 (*BBS20*)
IFT27 (*BBS19*)
IFT74 (*BBS22*)
INPP5E
ISL1
KIDINS220
KSR2
LEP
LEPR
LZTFL1 (*BBS17*)
MAGEL2
MC3R
MC4R
MECP2
MKKS (*BBS6*)
MKS1 (*BBS13*)
MRAP2
NCOA1 (*SRC1*)
NR0B2
NRP1
NRP2
NTRK2
PCNT
PCSK1
PHF6
PHIP
PLXNA1
PLXNA2
PLXNA3
PLXNA4
POMC
PPARG
PROK2
RAB23
RAI1
RPGRIP1L
RPS6KA3
SDCCAG8 (*BBS16*)
SEMA3A
SEMA3B
SEMA3C
SEMA3D
SEMA3E
SEMA3F
SEMA3G
SH2B1
SIM1
TBX3
TRIM32 (*BBS11*)
TRPC5
TTC8 (*BBS8*)
TUB
UCP3
VPS13B
WDPCP (*BBS15*)
16p11.2 chromosomal region

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