



Uncovering Rare Obesity® Gene Panel

The Uncovering Rare Obesity test panel includes 87 genes (including 29 BBS-associated genes) and 1 chromosomal region, reflective of nearly all of the most frequently tested genes associated with obesity.

Gene Symbol or Region (alternate*)	OMIM Gene Number	Gene Symbol or Region (alternate*)	OMIM Gene Number	Bardet-Biedl syndrome-associated genes:	
				Gene Symbol or Region (alternate*)	OMIM Gene Number
ADCY3	600291	PCNT	605925	ARL6 (BBS3)	608845
AFF4	604417	PCSK1	162150	BBIP1 (BBS18)	613605
ALMS1	606844	PHF6	300414	BBS1	209901
ASIP	600201	PHIP	612870	BBS2	606151
BDNF	113505	PLXNA1	601055	BBS4	600374
CPE	114855	PLXNA2	601054	BBS5	603650
CREBBP	600140	PLXNA3	300022	BBS7	607590
CUL4B	300304	PLXNA4	604280	BBS9 (PTHB1)	607968
DNMT3A	602769	POMC	176830	BBS10	610148
DYRK1B	604556	PPARG	601487	BBS12	610683
EP300	602700	PROK2	607002	CEP164	614848
GNAS	139320	RAB23	606144	CEP290 (BBS14)	610142
HTR2C	312861	RAI1	607642	CFAP418 (BBS21)	614477
INPP5E	613037	RPGRIP1L	610937	IFT27 (BBS19)	615870
ISL1	600366	RPS6KA3	300075	IFT74 (BBS22)	608040
KIDINS220	615759	SEMA3A	603961	IFT172 (BBS20)	607386
KSR2	610737	SEMA3B	601281	LRR45	
LEP	164160	SEMA3C	602645	LZTFL1 (BBS17)	606568
LEPR	601007	SEMA3D	609907	MKKS (BBS6)	604896
MAGEL2	605283	SEMA3E	608166	MKS1 (BBS13)	609883
MC3R	155540	SEMA3F	601124	NPHP1	607100
MC4R	155541	SEMA3G	620997	SCAPER	611611
MECP2	300005	SH2B1	608937	SCLT1	611399
MRAP2	615410	SIM1	603128	SDCCAG8 (BBS16)	613524
NCOA1 (SRC1)	602691	TBX3	601621	TMEM67	609884
NR0B2	604630	TRPC5	300334	TRIM32 (BBS11)	602290
NRP1	602069	TUB	601197	TTC8 (BBS8)	608132
NRP2	602070	UCP3	602044	TTC21B	612014
NTRK2	600456	VPS13B	607817	WDPCP (BBS15)	613580
		16p11.2 chromosomal region†	613444‡		

*Alternate gene symbols or phenotype abbreviations listed for clarity

†Assessment for rearrangement of the 16p11.2 chromosomal region

‡OMIM phenotype number

Gene symbols and alternative names may change and are current as of September 2025.

Questions? Contact PreventionGenetics

3800 South Business Park Avenue, Marshfield, Wisconsin 54449 • Phone: 1-844-513-3994 • Fax: 715-384-3661 • Email: support@preventiongenetics.com

© 2025, Rhythm Pharmaceuticals, Inc. All rights reserved. Rhythm, Uncovering Rare Obesity, and their logos are trademarks of Rhythm Pharmaceuticals, Inc. MD-US-NP-2000005 (11.09/2025)