



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 | BBS10 | 610148 |
| ASIP | 600201 | PHIP | 612870 | BBS12 | 610683 |
| BDNF | 113505 | PLXNA1 | 601055 | BBS1 | 209901 |
| CPE | 114855 | PLXNA2 | 601054 | BBS2 | 606151 |
| CREBBP | 600140 | PLXNA3 | 300022 | BBS4 | 600374 |
| CUL4B | 300304 | PLXNA4 | 604280 | BBS5 | 603650 |
| DNMT3A | 602769 | POMC | 176830 | BBS7 | 607590 |
| DYRK1B | 604556 | PPARG | 601487 | BBS9 (PTHB1) | 607968 |
| EP300 | 602700 | PROK2 | 607002 | CFAP418 (BBS21) | 614477 |
| GNAS | 139320 | RAB23 | 606144 | CEP164 | 614848 |
| HTR2C | 312861 | RAI1 | 607642 | CEP290 (BBS14) | 610142 |
| INPP5E | 613037 | RPGRIP1L | 610937 | IFT172 (BBS20) | 607386 |
| ISL1 | 600366 | RPS6KA3 | 300075 | IFT27 (BBS19) | 615870 |
| KIDINS220 | 615759 | SEMA3A | 603961 | IFT74 (BBS22) | 608040 |
| 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 | | 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† | | | |

*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 2024].