

|                 |                                                                         |
|-----------------|-------------------------------------------------------------------------|
| Full name:      | Lipodystrophy and/or hyperinsulinism (30 genes) - IPG                   |
| Type of panel:  | <u>Custom panel</u>                                                     |
| Provider:       | home made                                                               |
| Version number: | V7                                                                      |
| Laboratory:     | <u>Centre de Génétique-Institut de Pathologie et de Génétique (IPG)</u> |
| Created:        | 14 Dec 2022 - 14:26                                                     |
| Changed:        | 11 Dec 2023 - 12:55                                                     |

## Related Analytes

| GENE          | % OF CODING SEQUENCE SUFFICIENTLY COVERED TO<br>DETECT HETEROZYGOUS MUTATIONS | COPY NUMBER<br>VARIATION | COMMENTS       |
|---------------|-------------------------------------------------------------------------------|--------------------------|----------------|
| <u>ABCC8</u>  | 100.00                                                                        | 1                        | NM_000352.6    |
| <u>AGPAT2</u> | 100.00                                                                        | 1                        | NM_006412.4    |
| <u>AKT2</u>   | 100.00                                                                        | 1                        | NM_001626.6    |
| <u>ALMS1</u>  | 100.00                                                                        | 1                        | NM_001378454.1 |
| <u>BLM</u>    | 100.00                                                                        | 1                        | NM_000057.4    |
| <u>BSCL2</u>  | 100.00                                                                        | 1                        | NM_001122955.4 |
| <u>CAV1</u>   | 100.00                                                                        | 1                        | NM_001753.5    |
| <u>CAVIN1</u> | 100.00                                                                        | 1                        | NM_012232.6    |
| <u>CIDEC</u>  | 100.00                                                                        | 1                        | NM_001321142.2 |
| <u>GCK</u>    | 100.00                                                                        | 1                        | NM_000162.5    |
| <u>GLUD1</u>  | 100.00                                                                        | 1                        | NM_005271.5    |

| GENE            | % OF CODING SEQUENCE SUFFICIENTLY COVERED TO<br>DETECT HETEROZYGOUS MUTATIONS | COPY NUMBER<br>VARIATION | COMMENTS       |
|-----------------|-------------------------------------------------------------------------------|--------------------------|----------------|
| <u>HADH</u>     | 100.00                                                                        | 1                        | NM_005327.7    |
| <u>HNF1A</u>    | 100.00                                                                        | 1                        | NM_000545.8    |
| <u>HNF4A</u>    | 100.00                                                                        | 1                        | NM_175914.5    |
| <u>INSR</u>     | 100.00                                                                        | 1                        | NM_000208.4    |
| <u>KCNJ11</u>   | 100.00                                                                        | 1                        | NM_000525.4    |
| <u>LIPE</u>     | 100.00                                                                        | 1                        | NM_005357.4    |
| <u>LMNA</u>     | 100.00                                                                        | 1                        | NM_170707.4    |
| <u>MAFA</u>     | 100.00                                                                        | 1                        | NM_201589.4    |
| <u>PCNT</u>     | 100.00                                                                        | 1                        | NM_006031.6    |
| <u>PCYT1A</u>   | 100.00                                                                        | 1                        | NM_001312673.2 |
| <u>PIK3R1</u>   | 100.00                                                                        | 1                        | NM_181523.3    |
| <u>PLAAT3</u>   | 100.00                                                                        | 1                        | NM_001128203.2 |
| <u>POLD1</u>    | 100.00                                                                        | 1                        | NM_002691.4    |
| <u>PPARG</u>    | 100.00                                                                        | 1                        | NM_138711.6    |
| <u>PPP1R3A</u>  | 100.00                                                                        | 1                        | NM_002711.4    |
| <u>SLC16A1</u>  | 100.00                                                                        | 1                        | NM_003051.4    |
| <u>UCP2</u>     | 100.00                                                                        | 1                        | NM_003355.3    |
| <u>WRN</u>      | 100.00                                                                        | 1                        | NM_000553.6    |
| <u>ZMPSTE24</u> | 100.00                                                                        | 1                        | NM_005857.5    |