

|                 |                                                                                                                       |
|-----------------|-----------------------------------------------------------------------------------------------------------------------|
| Full name:      | Myopathy (genepanel) - UZA                                                                                            |
| Description:    | <a href="https://labogidsmedgen.uza.be/analyses/myopathie-mu">https://labogidsmedgen.uza.be/analyses/myopathie-mu</a> |
| Version number: | MU_versie 3                                                                                                           |
| Laboratory:     | <a href="#">Centrum Medische Genetica - UZ Antwerpen</a>                                                              |
| Created:        | 11 Dec 2020 - 08:57                                                                                                   |
| Changed:        | 27 Nov 2023 - 09:19                                                                                                   |

Related Analytes

| GENE                   | % OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS | COPY NUMBER VARIATION | COMMENTS |
|------------------------|----------------------------------------------------------------------------|-----------------------|----------|
| <a href="#">ABHD5</a>  | 99.64                                                                      | 1                     |          |
| <a href="#">ACAD9</a>  | 99.97                                                                      | 1                     |          |
| <a href="#">ACADM</a>  | 99.64                                                                      | 1                     |          |
| <a href="#">ACADS</a>  | 100.00                                                                     | 1                     |          |
| <a href="#">ACADVL</a> | 99.94                                                                      | 1                     |          |
| <a href="#">ACTA1</a>  | 99.99                                                                      | 1                     |          |
| <a href="#">ACTN2</a>  | 99.96                                                                      | 1                     |          |
| <a href="#">ADSS1</a>  | 99.99                                                                      | 1                     |          |
| <a href="#">AGK</a>    | 99.98                                                                      | 1                     |          |
| <a href="#">AGL</a>    | 99.92                                                                      | 1                     |          |
| <a href="#">AGRN</a>   | 99.83                                                                      | 1                     |          |
| <a href="#">ALDOA</a>  | 100.00                                                                     | 1                     |          |

| GENE            | % OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT<br>HETEROZYGOUS MUTATIONS | COPY NUMBER<br>VARIATION | COMMENTS |
|-----------------|-------------------------------------------------------------------------------|--------------------------|----------|
| <u>ALG14</u>    | 99.97                                                                         | 1                        |          |
| <u>ALG2</u>     | 99.95                                                                         | 1                        |          |
| <u>ANO5</u>     | 99.96                                                                         | 1                        |          |
| <u>ANXA11</u>   | 99.88                                                                         | 1                        |          |
| <u>ASCC1</u>    | 91.38                                                                         | 1                        |          |
| <u>ATP1A2</u>   | 99.96                                                                         | 1                        |          |
| <u>ATP2A1</u>   | 99.97                                                                         | 1                        |          |
| <u>B3GALNT2</u> | 94.03                                                                         | 1                        |          |
| <u>B4GAT1</u>   | 99.95                                                                         | 1                        |          |
| <u>BAG3</u>     | 100.00                                                                        | 1                        |          |
| <u>BIN1</u>     | 100.00                                                                        | 1                        |          |
| <u>BVES</u>     | 99.97                                                                         | 1                        |          |
| <u>CACNA1S</u>  | 99.92                                                                         | 1                        |          |
| <u>CAP2</u>     | 99.97                                                                         | 1                        |          |
| <u>CAPN3</u>    | 99.98                                                                         | 1                        |          |
| <u>CASQ1</u>    | 99.91                                                                         | 1                        |          |
| <u>CAV3</u>     | 99.85                                                                         | 1                        |          |
| <u>CAVIN1</u>   | 99.99                                                                         | 1                        |          |
| <u>CFL2</u>     | 99.81                                                                         | 1                        |          |
| <u>CHAT</u>     | 99.77                                                                         | 1                        |          |

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|---------------|-------------------------------------------------------------------------------|--------------------------|----------|
| <u>CHD7</u>   | 99.96                                                                         | 1                        |          |
| <u>CHKB</u>   | 99.89                                                                         | 1                        |          |
| <u>CHRNA1</u> | 99.96                                                                         | 1                        |          |
| <u>CHRNA1</u> | 99.99                                                                         | 1                        |          |
| <u>CHRNA1</u> | 100.00                                                                        | 1                        |          |
| <u>CHRNA1</u> | 99.95                                                                         | 1                        |          |
| <u>CHRNA1</u> | 99.87                                                                         | 1                        |          |
| <u>CHRNA1</u> | 99.97                                                                         | 1                        |          |
| <u>CHRNA1</u> | 99.97                                                                         | 1                        |          |
| <u>CHRNA1</u> | 99.95                                                                         | 1                        |          |
| <u>CHRNA1</u> | 99.56                                                                         | 1                        |          |
| <u>CHRNA1</u> | 99.96                                                                         | 1                        |          |
| <u>CHRNA1</u> | 100.00                                                                        | 1                        |          |
| <u>CHRNA1</u> | 99.96                                                                         | 1                        |          |
| <u>CHRNA1</u> | 99.99                                                                         | 1                        |          |
| <u>CHRNA1</u> | 99.97                                                                         | 1                        |          |
| <u>CHRNA1</u> | 98.51                                                                         | 1                        |          |
| <u>CHRNA1</u> | 99.99                                                                         | 1                        |          |
| <u>CHRNA1</u> | 99.59                                                                         | 1                        |          |
| <u>CHRNA1</u> | 99.98                                                                         | 1                        |          |

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|----------------|-------------------------------------------------------------------------------|--------------------------|----------|
| <u>DAG1</u>    | 100.00                                                                        | 1                        |          |
| <u>DCST2</u>   | 99.92                                                                         | 1                        |          |
| <u>DES</u>     | 100.00                                                                        | 1                        |          |
| <u>DGUOK</u>   | 99.76                                                                         | 1                        |          |
| <u>DMD</u>     | 99.41                                                                         | 1                        |          |
| <u>DMPK</u>    | 99.98                                                                         | 1                        |          |
| <u>DNAJB6</u>  | 99.91                                                                         | 1                        |          |
| <u>DNM2</u>    | 99.96                                                                         | 1                        |          |
| <u>DNMT3B</u>  | 99.98                                                                         | 1                        |          |
| <u>DOK7</u>    | 99.73                                                                         | 1                        |          |
| <u>DPAGT1</u>  | 99.96                                                                         | 1                        |          |
| <u>DPM2</u>    | 100.00                                                                        | 1                        |          |
| <u>DPM3</u>    | 100.00                                                                        | 1                        |          |
| <u>DYSF</u>    | 99.99                                                                         | 1                        |          |
| <u>EMD</u>     | 97.92                                                                         | 1                        |          |
| <u>ENO3</u>    | 99.98                                                                         | 1                        |          |
| <u>ETFA</u>    | 99.84                                                                         | 1                        |          |
| <u>ETFDH</u>   | 99.76                                                                         | 1                        |          |
| <u>FAM111B</u> | 99.99                                                                         | 1                        |          |
| <u>FDX2</u>    | 99.94                                                                         | 1                        |          |

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|----------------|-------------------------------------------------------------------------------|--------------------------|----------|
| <u>FHL1</u>    | 99.92                                                                         | 1                        |          |
| <u>FKRP</u>    | 99.99                                                                         | 1                        |          |
| <u>FKTN</u>    | 99.91                                                                         | 1                        |          |
| <u>FLAD1</u>   | 100.00                                                                        | 1                        |          |
| <u>FLNC</u>    | 99.99                                                                         | 1                        |          |
| <u>FXR1</u>    | 99.81                                                                         | 1                        |          |
| <u>GAA</u>     | 99.99                                                                         | 1                        |          |
| <u>GBE1</u>    | 99.84                                                                         | 1                        |          |
| <u>GFER</u>    | 99.96                                                                         | 1                        |          |
| <u>GFPT1</u>   | 99.90                                                                         | 1                        |          |
| <u>GGPS1</u>   | 99.89                                                                         | 1                        |          |
| <u>GMPPB</u>   | 99.95                                                                         | 1                        |          |
| <u>GNE</u>     | 99.98                                                                         | 1                        |          |
| <u>GYG1</u>    | 99.97                                                                         | 1                        |          |
| <u>GYS1</u>    | 99.98                                                                         | 1                        |          |
| <u>HACD1</u>   | 99.78                                                                         | 1                        |          |
| <u>HADH</u>    | 99.97                                                                         | 1                        |          |
| <u>HADHA</u>   | 99.96                                                                         | 1                        |          |
| <u>HADHB</u>   | 99.97                                                                         | 1                        |          |
| <u>HNRNPA1</u> | 100.00                                                                        | 1                        |          |

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|------------------|-------------------------------------------------------------------------------|--------------------------|----------|
| <u>HNRNPA2B1</u> | 99.95                                                                         | 1                        |          |
| <u>HNRNPDL</u>   | 99.92                                                                         | 1                        |          |
| <u>INPP5K</u>    | 99.99                                                                         | 1                        |          |
| <u>ISCU</u>      | 99.94                                                                         | 1                        |          |
| <u>ITGA7</u>     | 99.98                                                                         | 1                        |          |
| <u>ITGA9</u>     | 99.80                                                                         | 1                        |          |
| <u>JAG2</u>      | 98.50                                                                         | 1                        |          |
| <u>KBTBD13</u>   | 100.00                                                                        | 1                        |          |
| <u>KCNJ2</u>     | 100.00                                                                        | 1                        |          |
| <u>KIF21A</u>    | 99.80                                                                         | 1                        |          |
| <u>KLHL40</u>    | 100.00                                                                        | 1                        |          |
| <u>KLHL41</u>    | 99.96                                                                         | 1                        |          |
| <u>KLHL9</u>     | 100.00                                                                        | 1                        |          |
| <u>KY</u>        | 98.80                                                                         | 1                        |          |
| <u>LAMA2</u>     | 99.97                                                                         | 1                        |          |
| <u>LAMB2</u>     | 99.99                                                                         | 1                        |          |
| <u>LAMP2</u>     | 98.98                                                                         | 1                        |          |
| <u>LARGE1</u>    | 99.99                                                                         | 1                        |          |
| <u>LDB3</u>      | 98.02                                                                         | 1                        |          |
| <u>LDHA</u>      | 99.77                                                                         | 1                        |          |

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|----------------|-------------------------------------------------------------------------------|--------------------------|----------|
| <u>LIMS2</u>   | 99.65                                                                         | 1                        |          |
| <u>LMNA</u>    | 99.98                                                                         | 1                        |          |
| <u>LMOD3</u>   | 99.96                                                                         | 1                        |          |
| <u>LPIN1</u>   | 99.97                                                                         | 1                        |          |
| <u>LRP4</u>    | 99.98                                                                         | 1                        |          |
| <u>MAP3K20</u> | 99.92                                                                         | 1                        |          |
| <u>MATR3</u>   | 99.87                                                                         | 1                        |          |
| <u>MB</u>      | 99.99                                                                         | 1                        |          |
| <u>MEGF10</u>  | 99.90                                                                         | 1                        |          |
| <u>MICU1</u>   | 99.98                                                                         | 1                        |          |
| <u>MLIP</u>    | 99.94                                                                         | 1                        |          |
| <u>MSTO1</u>   | 98.87                                                                         | 1                        |          |
| <u>MTM1</u>    | 99.36                                                                         | 1                        |          |
| <u>MUSK</u>    | 99.97                                                                         | 1                        |          |
| <u>MYBPC1</u>  | 99.90                                                                         | 1                        |          |
| <u>MYBPC3</u>  | 99.98                                                                         | 1                        |          |
| <u>MYH14</u>   | 99.79                                                                         | 1                        |          |
| <u>MYH2</u>    | 99.97                                                                         | 1                        |          |
| <u>MYH3</u>    | 99.96                                                                         | 1                        |          |
| <u>MYH7</u>    | 100.00                                                                        | 1                        |          |

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|---------------|-------------------------------------------------------------------------------|--------------------------|----------|
| <u>MYL1</u>   | 99.93                                                                         | 1                        |          |
| <u>MYL2</u>   | 99.98                                                                         | 1                        |          |
| <u>MYMX</u>   | 100.00                                                                        | 1                        |          |
| <u>MYO18B</u> | 99.98                                                                         | 1                        |          |
| <u>MYO9A</u>  | 99.91                                                                         | 1                        |          |
| <u>MYOD1</u>  | 99.82                                                                         | 1                        |          |
| <u>MYOT</u>   | 99.98                                                                         | 1                        |          |
| <u>MYPN</u>   | 99.97                                                                         | 1                        |          |
| <u>NEB</u>    | 99.77                                                                         | 1                        |          |
| <u>NPL</u>    | 99.95                                                                         | 1                        |          |
| <u>OPA1</u>   | 99.87                                                                         | 1                        |          |
| <u>ORAI1</u>  | 97.73                                                                         | 1                        |          |
| <u>PABPN1</u> | 99.67                                                                         | 1                        |          |
| <u>PAX7</u>   | 99.96                                                                         | 1                        |          |
| <u>PFKM</u>   | 99.82                                                                         | 1                        |          |
| <u>PGAM2</u>  | 100.00                                                                        | 1                        |          |
| <u>PGK1</u>   | 99.71                                                                         | 1                        |          |
| <u>PGM1</u>   | 99.89                                                                         | 1                        |          |
| <u>PHKA1</u>  | 99.57                                                                         | 1                        |          |
| <u>PHKB</u>   | 99.79                                                                         | 1                        |          |



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|----------------|-------------------------------------------------------------------------------|--------------------------|----------|
| <u>PHOX2A</u>  | 96.75                                                                         | 1                        |          |
| <u>PLEC</u>    | 99.98                                                                         | 1                        |          |
| <u>PNPLA2</u>  | 99.99                                                                         | 1                        |          |
| <u>PNPLA8</u>  | 99.81                                                                         | 1                        |          |
| <u>POGLUT1</u> | 99.91                                                                         | 1                        |          |
| <u>POLG</u>    | 99.98                                                                         | 1                        |          |
| <u>POLG2</u>   | 99.68                                                                         | 1                        |          |
| <u>POMGNT1</u> | 99.98                                                                         | 1                        |          |
| <u>POMGNT2</u> | 100.00                                                                        | 1                        |          |
| <u>POMK</u>    | 99.98                                                                         | 1                        |          |
| <u>POMT1</u>   | 99.91                                                                         | 1                        |          |
| <u>POMT2</u>   | 99.93                                                                         | 1                        |          |
| <u>POPDC3</u>  | 99.93                                                                         | 1                        |          |
| <u>PREPL</u>   | 99.96                                                                         | 1                        |          |
| <u>PRKAG2</u>  | 99.92                                                                         | 1                        |          |
| <u>PYGM</u>    | 99.98                                                                         | 1                        |          |
| <u>PYROXD1</u> | 99.83                                                                         | 1                        |          |
| <u>RAPSN</u>   | 99.99                                                                         | 1                        |          |
| <u>RBCK1</u>   | 99.99                                                                         | 1                        |          |
| <u>RXYLT1</u>  | 99.53                                                                         | 1                        |          |

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|-----------------|-------------------------------------------------------------------------------|--------------------------|----------|
| <u>RYR1</u>     | 99.92                                                                         | 1                        |          |
| <u>SCN4A</u>    | 99.77                                                                         | 1                        |          |
| <u>SELENON</u>  | 95.81                                                                         | 1                        |          |
| <u>SGCA</u>     | 99.99                                                                         | 1                        |          |
| <u>SGCB</u>     | 99.62                                                                         | 1                        |          |
| <u>SGCD</u>     | 99.94                                                                         | 1                        |          |
| <u>SGCG</u>     | 99.87                                                                         | 1                        |          |
| <u>SIL1</u>     | 99.99                                                                         | 1                        |          |
| <u>SLC18A3</u>  | 100.00                                                                        | 1                        |          |
| <u>SLC22A5</u>  | 99.99                                                                         | 1                        |          |
| <u>SLC25A20</u> | 99.90                                                                         | 1                        |          |
| <u>SLC25A4</u>  | 99.96                                                                         | 1                        |          |
| <u>SLC5A7</u>   | 99.97                                                                         | 1                        |          |
| <u>SMCHD1</u>   | 99.82                                                                         | 1                        |          |
| <u>SMPX</u>     | 99.37                                                                         | 1                        |          |
| <u>SPEG</u>     | 99.97                                                                         | 1                        |          |
| <u>SQSTM1</u>   | 100.00                                                                        | 1                        |          |
| <u>STAC3</u>    | 99.94                                                                         | 1                        |          |
| <u>STIM1</u>    | 99.97                                                                         | 1                        |          |
| <u>SVIL</u>     | 99.94                                                                         | 1                        |          |

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|-----------------|-------------------------------------------------------------------------------|--------------------------|----------|
| <u>SYNE1</u>    | 99.96                                                                         | 1                        |          |
| <u>SYNE2</u>    | 99.85                                                                         | 1                        |          |
| <u>SYT2</u>     | 99.89                                                                         | 1                        |          |
| <u>TCAP</u>     | 100.00                                                                        | 1                        |          |
| <u>TIA1</u>     | 99.56                                                                         | 1                        |          |
| <u>TK2</u>      | 99.99                                                                         | 1                        |          |
| <u>TMEM126B</u> | 99.68                                                                         | 1                        |          |
| <u>TMEM43</u>   | 99.98                                                                         | 1                        |          |
| <u>TNNC2</u>    | 99.95                                                                         | 1                        |          |
| <u>TNNI2</u>    | 100.00                                                                        | 1                        |          |
| <u>TNNT1</u>    | 99.82                                                                         | 1                        |          |
| <u>TNNT3</u>    | 100.00                                                                        | 1                        |          |
| <u>TNPO3</u>    | 99.95                                                                         | 1                        |          |
| <u>TNXB</u>     | 99.95                                                                         | 1                        |          |
| <u>TOP3A</u>    | 99.95                                                                         | 1                        |          |
| <u>TOR1AIP1</u> | 99.84                                                                         | 1                        |          |
| <u>TPI1</u>     | 99.95                                                                         | 1                        |          |
| <u>TPM2</u>     | 99.93                                                                         | 1                        |          |
| <u>TPM3</u>     | 99.95                                                                         | 1                        |          |
| <u>TRAPPC11</u> | 99.59                                                                         | 1                        |          |

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|---------------|-------------------------------------------------------------------------------|--------------------------|----------|
| <u>TRDN</u>   | 99.03                                                                         | 1                        |          |
| <u>TRIM32</u> | 100.00                                                                        | 1                        |          |
| <u>TRIP4</u>  | 99.91                                                                         | 1                        |          |
| <u>TRMT5</u>  | 99.83                                                                         | 1                        |          |
| <u>TTN</u>    | 99.96                                                                         | 1                        |          |
| <u>TUBB3</u>  | 100.00                                                                        | 1                        |          |
| <u>TWINK</u>  | 100.00                                                                        | 1                        |          |
| <u>UNC45B</u> | 99.93                                                                         | 1                        |          |
| <u>VAMP1</u>  | 99.70                                                                         | 1                        |          |
| <u>VCP</u>    | 99.92                                                                         | 1                        |          |
| <u>VMA21</u>  | 97.04                                                                         | 1                        |          |
| <u>VWA1</u>   | 91.35                                                                         | 1                        |          |