## Huntington disease Reference Materials Characterized by the GeT-RM

## CAG Repeat Length as Determined by:

10 Clinical Genetic Laboratories
Mean

| Allele 1 (95\% confidence interval) | Allele 2 (95\% confidence interval) | Allele 1 (95\% confidence interval) | Allele 2 (95\% confidence interval) | Allele 1/2 | Cell Bank | DNA <br> Number | Characterization methods ${ }^{4}$ (\# labs) |
| :---: | :---: | :---: | :---: | :---: | :---: | :---: | :---: |
| $\begin{gathered} 15 \\ (13.7-15.8) \\ \hline \end{gathered}$ | $\begin{gathered} 15 \\ (13.7-15.8) \\ \hline \end{gathered}$ | $\begin{gathered} 15 \\ (77 \%) \end{gathered}$ | $\begin{gathered} \hline 15 \\ (80 \%) \\ \hline \end{gathered}$ | 15/15 | Coriell | NA20245 | PCR-PAGE (5), PCR-CE (4), PCR-seq (1) |
| $\begin{gathered} 17 \\ (15.9-18.3) \end{gathered}$ | $\begin{gathered} 18 \\ (17.2-19.1) \end{gathered}$ | $\begin{gathered} 17 \\ (77 \%) \\ \hline \end{gathered}$ | $\begin{gathered} 18 \\ (87 \%) \\ \hline \end{gathered}$ | 17/18 | Coriell | NA20206 | $\begin{gathered} \hline \text { PCR-PAGE (5), PCR-CE (4), } \\ \text { PCR-seq (1) } \end{gathered}$ |
| $\begin{gathered} 19 \\ (17.7-20.7) \end{gathered}$ | $\begin{gathered} 21 \\ (20-22.1) \end{gathered}$ | $\begin{gathered} 19 \\ (67 \%) \end{gathered}$ | $\begin{gathered} 21 \\ (77 \%) \end{gathered}$ | 19/21 | Coriell | NA20207 | $\begin{gathered} \hline \text { PCR-PAGE (5), PCR-CE (4), } \\ \text { PCR-seq (1) } \end{gathered}$ |
| $\begin{gathered} 15 \\ (14.1-15.9) \end{gathered}$ | $\begin{gathered} 24 \\ (22.3-25.9) \\ \hline \end{gathered}$ | $\begin{gathered} 15 \\ (80 \%) \end{gathered}$ | $\begin{gathered} 24 \\ (77 \%) \\ \hline \end{gathered}$ | 15/24 | Coriell | NA20246 | PCR-PAGE (5), PCR-CE (4), PCR-seq (1) |
| $\begin{gathered} 15 \\ (14.1-15.9) \end{gathered}$ | $\begin{gathered} 29 \\ (28.1-29.9) \\ \hline \end{gathered}$ | $\begin{gathered} 15 \\ (80 \%) \end{gathered}$ | $\begin{gathered} 29 \\ (80 \%) \end{gathered}$ | 15/29 | Coriell | NA20247 | $\begin{gathered} \hline \text { PCR-PAGE (5), PCR-CE (4), } \\ \text { PCR-seq (1) } \end{gathered}$ |
| $\begin{gathered} 17 \\ (15.0-19.3) \end{gathered}$ | $\begin{gathered} 36 \\ (35.1-37.3) \\ \hline \end{gathered}$ | $\begin{gathered} 17 \\ (77 \%) \end{gathered}$ | $\begin{gathered} 36 \\ (80 \%) \end{gathered}$ | 17/36 | Coriell | NA20248 | $\begin{gathered} \hline \text { PCR-PAGE (5), PCR-CE (4), } \\ \text { PCR-seq (1) } \end{gathered}$ |
| $\begin{gathered} 22 \\ (21.1-22.8) \end{gathered}$ | $\begin{gathered} 39 \\ (38.3-39.9) \\ \hline \end{gathered}$ | $\begin{gathered} 22 \\ (87 \%) \\ \hline \end{gathered}$ | $\begin{gathered} 39 \\ (83 \%) \\ \hline \end{gathered}$ | 22/39 | Coriell | NA20249 | $\begin{gathered} \hline \text { PCR-PAGE (5), PCR-CE (4), } \\ \text { PCR-seq (1) } \end{gathered}$ |
| $\begin{gathered} 15 \\ (14.1-15.8) \end{gathered}$ | $\begin{gathered} 40 \\ (39.2-41.0) \end{gathered}$ | $\begin{gathered} 15 \\ (83 \%) \end{gathered}$ | $\begin{gathered} 40 \\ (80 \%) \end{gathered}$ | 15/40 | Coriell | NA20250 | $\begin{gathered} \hline \text { PCR-PAGE (5), PCR-CE (4), } \\ \text { PCR-seq (1) } \end{gathered}$ |
| $\begin{gathered} 35 \\ (33.4-36.3) \end{gathered}$ | $\begin{gathered} 45 \\ (43.5-46.5) \\ \hline \end{gathered}$ | $\begin{gathered} 35 \\ (80 \%) \end{gathered}$ | $\begin{gathered} 45 \\ (77 \%) \end{gathered}$ | 35/45 | Coriell | NA20208 | PCR-PAGE (5), PCR-CE (4), PCR-seq (1) |
| $\begin{gathered} 45 \\ (43.8-46.2) \\ \hline \end{gathered}$ | $\begin{gathered} 47 \\ (46.3-47.8) \\ \hline \end{gathered}$ | $\begin{gathered} 45 \\ (73 \%) \end{gathered}$ | $\begin{gathered} 47 \\ (83 \%) \\ \hline \end{gathered}$ | 45/46 | Coriell | NA20209 | $\begin{gathered} \hline \text { PCR-PAGE (5), PCR-CE (4), } \\ \text { PCR-seq (1) } \end{gathered}$ |
| $\begin{gathered} 39 \\ (38.1-40.0) \\ \hline \end{gathered}$ | $\begin{gathered} 50 \\ (49.1-50.8) \\ \hline \end{gathered}$ | $\begin{gathered} 39 \\ (80 \%) \end{gathered}$ | $\begin{gathered} 50 \\ (83 \%) \\ \hline \end{gathered}$ | 39/50 | Coriell | NA20251 | $\begin{gathered} \hline \text { PCR-PAGE (5), PCR-CE (4), } \\ \text { PCR-seq (1) } \\ \hline \end{gathered}$ |
| $\begin{gathered} 22 \\ (21.2-22.9) \\ \hline \end{gathered}$ | $\begin{gathered} 66 \\ (63.7-67.5) \end{gathered}$ | $\begin{gathered} 22 \\ (83 \%) \end{gathered}$ | $\begin{aligned} & 65 \text { (37\%) } \\ & 66 \text { (37\%) } \\ & \hline \end{aligned}$ | 22/65 | Coriell | NA20252 | $\begin{gathered} \hline \text { PCR-PAGE (5), PCR-CE (4), } \\ \text { PCR-seq (1) } \end{gathered}$ |
| $\begin{gathered} 17 \\ (15.4-18.2) \end{gathered}$ | $\begin{gathered} 74 \\ (72.0-76.6) \end{gathered}$ | $\begin{gathered} 17 \\ (77 \%) \end{gathered}$ | $\begin{gathered} 74 \\ (50 \%) \end{gathered}$ | $17 / 75$ | Coriell | NA20210 | $\begin{gathered} \hline \text { PCR-PAGE (5), PCR-CE (4), } \\ \text { PCR-seq (1) } \end{gathered}$ |
| $\begin{gathered} 22 \\ (20.3-23.3) \\ \hline \end{gathered}$ | $\begin{gathered} 99 \\ (95.8-102.7) \end{gathered}$ | $\begin{gathered} 22 \\ (73 \%) \end{gathered}$ | $\begin{gathered} 100 \\ (52 \%) \end{gathered}$ | 22/101 | Coriell | NA20253 | $\begin{gathered} \hline \text { PCR-PAGE (5), PCR-CE (4), } \\ \text { PCR-seq (1) } \\ \hline \end{gathered}$ |
|  |  |  |  |  |  |  | last updated 02-25-2008 |

${ }^{1}$ Mean repeat length calculated from 30 responses per allele (Except for allele 2 of DNA sample NA20253 which had only 25 responses). Values were rounded to the nearest whole number, ${ }^{2}$ CAG repeat length reported most often out of 30 responses per allele (Except for allele 2 of DNA sample NA20253 which had only 25 responses), ${ }^{3}$ Unidirectional sequencing, results from one lab (National Institute of Standards and Technology)
${ }^{4}$ Methods: All labs used home-brew methods. Each lab amplified the CAG repeat region using PCR and separated the products using either polyacrylmide gel electrophoresis (PAGE), an ALF automated sequencer (seq) or capillary electrophoresis (CE). PCR product sizing was done manually (PAGE) or was automated (CE + seq).

