



Domain KEY



N-terminal/Region I: Exons 2-17
 Central/Region II: Exons 39-46
 C-terminal/Region III: Exons 85-103

- Missense
- ◆ Nonsense
- Deletion
- Insertion
- △ Duplication
- * Splice Site

- MmD
- CCD
- Core Myopathy
- CNM/CNM-like
- CFTD
- RRM
- KDS
- Congenital Myopathy/MH
- AR MD
- Core/Rod Disease