**Supplemental file 3** Neuromuscular symptoms in patients referred because an *RYR1* variant was detected in diagnostic testing in the neuromuscular clinic without a specific clinical or histopathological diagnosis.

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| **Neuromuscular symptoms** | **n = 32** |
| (Exertional) myalgia and/or muscle cramps | 12 |
| (Exertional) myalgia or cramps in combination with muscle weakness | 5 |
| Muscle weakness, pattern of weakness not further specified | 4 |
| Proximal muscle weakness | 4 |
| Muscle weakness in combination with heat intolerance | 2 |
| (Exertional) myalgia or muscle cramps in combination with muscle stiffness | 1 |
| Necrotizing myopathy | 1 |
| Muscle stiffness | 1 |
| Exercise intolerance | 1 |
| (Exertional) myalgia or muscle cramps in combination with joint contractures | 1 |