**Appendix B: Questionnaire of survey**

***DEMOGRAPHICS***

1. **What was your sex assigned at birth?**
2. Male
3. Female
4. Prefer not to answer
5. **What gender do you identify as?**
6. Male
7. Female
8. Prefer not to answer
9. **What is your age?**
10. \_\_\_\_\_\_\_\_
11. Prefer not to answer

***DIAGNOSIS***

1. **How old were you when you were diagnosed with RYR-1-RD?**
2. \_\_\_\_\_\_\_\_
3. Prefer not to answer
4. **How was your RYR-1 related disease diagnosed?**
5. Muscle biopsy only
6. Genetic testing only
7. Both muscle biopsy and genetic testing
8. Assumed diagnosis based upon family history
9. Prefer not to answer
10. Other, please explain:
11. **What type of RYR-1-RD have you been diagnosed with?**
12. Central core disease (CCD)
13. Multi-minicore disease (MmD)
14. Centronuclear myopathy (CNM)
15. Congenital fiber type disproportion (CFTD)
16. Malignant hyperthermia susceptibility (MHS)
17. Prefer not to answer
18. Other, please explain:
19. **What is the inheritance of your RYR-1-RD?**
20. Autosomal dominant
21. Autosomal recessive
22. De Novo/Spontaneous
23. Unknown
24. Prefer not to answer
25. **What best describes your physical ability?**
26. Able to walk unassisted
27. Able to walk with assistance
28. Require wheelchair assistance
29. Require wheelchair
30. Prefer not to answer
31. **Do you think your RYR-1 related disease symptoms are progressive or non-progressive?**
32. Progressive (gotten worse)
33. Non-Progressive (stable)
34. Unsure
35. Prefer not to answer
36. Other, please explain:

***SYMPTOMS AND IMPACT OF THE CONDITION***

1. **Do you have any of the following signs and symptoms (please check all that apply)?**

* Muscle weakness
* Facial muscles and muscles of the arms and legs
* Delays in sitting, walking, etc.
* Difficulties getting up from the ﬂoor or out of a chair
* Difficulties walking or running
* Difficulties with stairs
* Fatigue, especially in warm weather
* Muscle wasting (muscles get smaller over time)
* Restricted movements of the eye muscles
* Fixed or stiff joints
* Changes in the shape of the chest
* Hip dislocation
* Eating difficulties
* Breathing difficulties, including sleep apnea
* Scoliosis
* Prefer not to answer
* Other, please explain:

1. **Does RYR-1-RD impact your well-being in any of the following areas (please check all that apply)?**
   1. Physical well-being
   2. Emotional well-being
   3. Nutritional well-being
   4. Social well-being
   5. Educational well-being
   6. Employment well-being
   7. Financial well-being
   8. Mental (intellectual) well-being
   9. Spiritual well-being
   10. Prefer not to answer
   11. Other, please explain:
2. **Please describe how your life is** **impacted (positively or negatively) to any of the well-being answers above.**
   1. Please explain:
   2. Prefer not to answer

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***PHYSICAL ACTIVITY***

1. **In a week, how many times on average do you engage in physical activity and/or exercise for more than 15 minutes?**
2. 1 – 2 times per week
3. 3 – 4 times per week
4. 5 – 6 times per wee
5. 7 or more times per week
6. I do not exercise
7. Prefer not to answer

**If you answered A thru D, please respond to next Questions 14 & 15. Otherwise, please respond see Question 16.**

1. **If you engage in physical activity and/or exercise, what types of activities do you do? Please check all that apply.**
2. Walking / wheelchair rolling
3. Bicycling
4. Dance
5. Physical therapy
6. Horseback riding
7. Swimming / water aerobics
8. Weight / resistance training
9. Activity Classes (i.e., pilates, yoga, or eccentrics)
10. Prefer not to answer
11. Other, please explain:
12. **If you exercise, have you been able to maintain or improve your strength and/or endurance?**
13. Yes
14. No
15. Not sure
16. Prefer not to answer
17. **I DO NOT engage in physical activity and/or exercise because (please check all that apply):**
18. I do not have time
19. I am concerned for my safety
20. I cannot design an exercise routine that works for me
21. I do not believe exercise will maintain or improve my condition
22. Prefer not to answer

***CLINICAL RESEARCH AND STUDIES***

1. **What is your willingness to participate in a clinical trial?**
2. I would **Definitely NOT** participate
3. I am **Unlikely** to participate
4. I am **Likely** to participate
5. I would **Definitely** participate
6. Prefer not to answer
7. **If you answered A. or B. in Question #17, Which below best explains why you do NOT or are UNLIKELY to participate in a clinical trial (please check all that apply)? Otherwise, please proceed to next Question.**
8. Concern for safety and tolerability
9. Too disruptive to life activities (i.e., school, work, family, personal life, etc.)
10. I do not believe there is hope for a treatment or cure
11. Prefer not to answer
12. Other, please explain:
13. **What do you think researchers need to know or better understand about living with RYR-1-RD?** 
    1. Prefer not to answer
    2. Please explain:
14. **In addition to the #1 goal of discovering a cure for RYR-1-RD, what aspects or areas of living with RYR-1 do you wish researchers would help make better?** 
    1. Prefer not to answer
    2. Please explain:
15. **What questions do you have for the medical community?**
16. How can I relieve everyday symptoms (i.e., muscle pain)?
17. How can I improve my physical well-being (i.e., fatigue, endurance)?
18. How can I improve my emotional well-being (i.e., anxiety)?
19. How can I improve my nutritional well-being (i.e., dietary supplements)?
20. What holistic approaches are beneficial? (i.e., Focusing on wellness and not just their illness or condition)?
21. How does the climate and environment impact my symptoms (i.e. .temperature)?