**APPENDIX B: Instruction for testimonials**

Our role in this workshop is to come together as patients/caregivers to represent the majority of the RYR-1 community. Each of you were strategically chosen to create a strong diverse group to ensure that we are representing as many individuals as we possibly can. Your patient story is your “voice.” It is a chance to really paint a picture of the life of an individual affected by RYR-1-related diseases (RYR-1-RD), and express what you would like to see change in medicine in the future. Our hope is that medical experts take this information as a reference when creating possible studies, treatments, and ultimately a cure. We are essentially saying “This is who we are, what we have been and are still going through, and what it will take for us to feel like we are content with the treatments and technologies available.”

**In respect to other presenters in the workshop, you will have about 7 minutes total to give your testimonial. Please time yourself prior to the workshop because we really want you to have enough time to express yourself.**

* Be open, honest, and clear.
* Share only what you’re comfortable sharing.
* Share the story of when and how you (or your loved one) were told that you have RYR-1 mutations. Not everyone’s journey starts at the same milestone. Some of us started at birth while others had their onset later in life. Our journeys are unique and that is what we want to emphasize.
* If you are a caregiver, please also talk about how this affects you as a caregiver.
* Supporting photos and short videos are strongly encouraged for your presentation. Visuals always help.
* If your RYR-1-RD began to affect you later in life, you may want to briefly share what your life was like prior and then focus on that pivotal moment that led to your diagnosis. What new symptoms did you experience? How has your work/social life changed?
* Talk about physical changes over time such as weakness and pain. What medications/supplements have and/or haven’t worked for you?
* Medical aids are commonly used for needs such as mobility, respiratory care, personal hygiene, and housekeeping. You may want to mention your aid such as a scooter, CPAP/BiPAP, grab bars, etc.
* You may want to talk about mental health, and how it may have an effect on you as well.
* Where would you like to see improvements regarding your expectations in medical research, studies, and treatment?
* This is your chance to ask thought provoking questions that you may have that your current doctor may not have an answer for.
* Sharing demographic information and diagnostic results may also be ideal.

You can view a couple of patient video stories including my own groundbreaking story at <https://www.ryr1.org/videos>, if you’d like some video inspiration. You’ll definitely see that we are all just as unique as we are alike, which is exactly why this group is so important.

You all are the stars of this workshop and are helping to provide key information for the future of RYR-1-RD. We want to thank you for your time and patience. Please do not hesitate to contact us should you have any further comments or questions about your personal stories.

Themes deduced from this instruction:

**DIAGNOSIS:** when and how was the RYR1-RM diagnosed?

* Life before the diagnosis
* Diagnosis

**SYMPTOMS AND IMPACT OF THE CONDITION**

* Symptoms
* Effect on physical functioning
* Effect on daily life
* Effect on mental health

**TREATMENT**

* Treatment

**EXPECTATIONS**

* Expectations
* Gaining knowledge