Appendix A: Interview Guide

Hello, I'm Fatima Amir from Cincinnati Children's Hospital Medical Center Division of Human Genetics. Could I speak with Mr. /Mrs. _____?

Thank you for your willingness to participate in this interview. This interview is part of an ongoing research study. The purpose of the study is to understand the clinical journey of patients diagnosed with a condition known by several names including Riboflavin Transporter Deficiency, Brown-Vialetto-Van Laere Syndrome or Fazio-Londe Syndrome. Which name are you most familiar with? _____ (*Depending on their answer, we will refer to the condition with the term that they are familiar with throughout the rest of the interview. For the purposes of this script, I will simply write it out as RTD*)

When we say we want to understand the clinical journey of patients diagnosed with *RTD*, we mean we want to understand the **order of their symptoms**, what **genetic testing** the patient has had until their final diagnosis of *RTD*, whether or not they have received any **incorrect diagnoses** before their diagnosis of *RTD* and their **healthcare interactions** – that is, what kinds of doctors they were seen by and in what order. We are mainly focusing on the journey up until the time of their genetic diagnosis with *RTD*.

During this interview, I will be recording responses as we go along so you may expect some pauses from my side after you answer a question.

To highlight some of the important elements of the consent, we would like to remind you that you are being asked to participate in this research because your child has a diagnosis of *RTD*. You should not participate in this research study if your child is over 18 years old and has not had a positive result for a mutation in the SLC52A2 gene. Your interview will take approximately one hour. Data collected from this research study will be maintained in a database that can be used for future studies on *RTD*. You will be asked for your permission to be involved in future research. You have the right to withdraw your information from the database at any time. About 20 people will take part in this study at Cincinnati Children's Hospital Medical Center. There are no known risks associated with the study. There may be unknown or unforeseen risks associated with study participation such as a loss of confidentiality. If you agree to take part in the research study, there is no direct medical benefit to you. The information learned from this study may benefit other patients with *RTD*. The information learned from this study may also help health care providers better understand the condition. It is hoped that this information will decrease the time to diagnosis, resulting in earlier treatment and a better prognosis for patients. Every effort will be made to maintain the confidentiality of your child's medical and research information. There are no costs to you for participation in this research study. There will be no payment or reimbursement to participate in this research study. If you refuse to participate, your rights concerning treatment, payment for services, and enrollment in a health plan or eligibility for benefits will not be affected.

We would like to remind you that your participation in this research study is completely voluntary and you may withdraw your consent at any time.

We would like to ask you some questions about your child's clinical journey. To help us better understand the more technical aspects of the journey, we may also like to review your child's medical records. We will ask your permission to access these records only if you have already sent these records to Dr. Keith Massey in the past, or if you were exclusively seen at Cincinnati Children's Hospital Medical Center. We will not ask you to collect and send us any records.

Would you like to participate in this study?

> **If no:** Thank you for your time.
>
> **If yes:** Thank you for volunteering to participate in our study. As the email explained, participating involves taking part in a phone interview and a possible review of your child's medical records. During the phone interview, we will ask you questions on your child's clinical journey till they received a diagnosis of *RTD*. For the medical records, if you have already sent your child's medical records to Dr. Keith Massey, then we can ask him to send them to us if you give us the permission to do so. If your child was seen exclusively at Cincinnati Children's, then we will ask you for permission to access your child's medical records.

I will now ask some demographic questions about your child.

What is the name of your child?

What is your relationship to *Patient*?

What is *Patient's* date of birth?

What is *Patient's* sex?

What is *Patient's* ethnicity?

Have you sent *Patient's* medical records to Dr. Keith Massey?

> **If yes:** Would you give us permission to ask Dr. Keith Massey for any of *Patient's* genetic testing results and audiology records up to the point of the final genetic testing confirming *Patient's* diagnosis?
>
> **If no:** Was *Patient* seen exclusively at Cincinnati Children's Hospital Medical Center?
>
> > **If yes:** We will mail you a release of information form that you can either mail back to us, or you can send us a scanned copy of, if you choose to allow us to access *Patient's* medical records directly.
> >
> > **If no:** We will not be conducting a medical record review for *Patient*.

At this time do you have any questions before we begin the interview?

> **If yes:** *Answer question as appropriate*
>
> **If no:** *Continue with interview.*

Was *Patient's* diagnosis made before confirming it with genetic testing, or was *Patient's* diagnosis made first with genetic testing?

If clinical dx first:

When was *Patient's* diagnosis of *RTD* made?

What kind of doctor made the diagnosis?

When was *Patient's* diagnosis of *RTD* confirmed with genetic testing?

If genetic dx first: When was genetic testing ordered that made a diagnosis of *RTD*? *Continue with interview.*

What kind of doctor ordered the genetic testing?

Do you know what kind of testing was done? Examples include Whole Exome Sequencing, a Gene Panel or Single gene testing for the SLC52A2 gene.

Thank you for that information. Now I will try to understand *Patient's* clinical journey until the diagnosis of *RTD* was made.

Going back in time a little bit, what was the first symptom that *Patient* developed?

What age was *he/she*? Or if you prefer to use dates, what month and year was that?

What doctor did you see at that time?

What was the outcome of that visit? Did they refer you to any other doctors?

What was the next symptom to develop?

(continue to ask and clarify order of symptoms and doctor visits up to the point of diagnosis)

I would like to summarize the information you have given me to make sure I have recorded it accurately. Please let me know if I have missed any information you deem relevant.

(Summarize information in table, and correct as appropriate)

(If genetic testing has not been mentioned) Was there any other genetic testing ordered by any other doctors along the way? What did it test for? Who ordered it? What were the results?

Were there any diagnoses that *Patient* was given that were incorrect before his/her correct diagnosis of *RTD*? Who made these diagnoses?

Now I will ask you some questions specifically related to Audiology.

What age was *Patient* identified with hearing loss *(if not already mentioned)*? Did *Patient* get hearing aids? Did *Patient* use cochlear implants? How well does *Patient* communicate now? How well do you think your child can hear with their devices?

Does *Patient* use speech to communicate? (*If yes, then do not ask remaining questions*)

Does *Patient* use sign language to communicate?

Does *Patient* use tactile signs to communicate?

Does *Patient* use a communication board to communicate?

Does *Patient* use gesturing to communicate?

Thank you. We value your opinion, so now I will ask you some general questions before we end the interview.

What research would you like to see done about *RTD* in the future?

Would you like to be contacted for any future research?

Do you have any other comments or feedback for us?

If records with Keith or CCHMC patient:

As mentioned earlier, we would also like to review some of *Patient's* medical records to get a more comprehensive picture.

If records with Keith: We will be getting in touch with Dr. Keith Massey and ask him to send us a copy of all of *Patient's* clinic notes, genetic testing results and audiology records that he has up to the time of diagnosis. Would you allow us to review these records?

If seen at CCHMC: Since your child was seen at Cincinnati Children's, we will be mailing you a release of information form in the next few days. If you give us permission to review your child's medical records, then you may sign and send the form back to us either by mail or by scanning or faxing it back to us. Then we will be able to access *Patient's* clinic notes, genetic testing results and audiology records up to the time of diagnosis. What mailing address can we use to mail the release of information form to you?

Thank you very much for agreeing to participate in this phone interview from the Division of Human Genetics at Cincinnati Children's Hospital Medical Center. Do you have any questions at this point?

I am the contact person for this study in case you have any questions that come up later. My contact information was included in the email previously sent to you regarding this study. If you

would like I can go over it again. Would you like me to review my contact information with you right now?

If yes:

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If no: *Continue with interview.*

Thank you again for your time. It was my pleasure speaking with you. Good bye!