Best Nursing Interventions for Educating Parents on Tyrosinemia Type 1

Julianna Louise Mize

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Best Nursing Interventions for Educating Parents on Tyrosinemia Type I

By

Julianna Louise Mize

An Honors Capstone

submitted in partial fulfillment of the requirements

for the Honors Certificate

to

The Honors College

Of

The University of Alabama in Huntsville

4/28/15

Honors Capstone Director: Ellise D. Adams, PhD, CNM

Associate Professor of Nursing

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Student (signature) Date

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Department Chair (signature) Date

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Honors College Dean (signature) Date
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Abstract

Tyrosinemia Type I is not a well-known disease among the general public. Since it is an autosomal recessive disorder, many carriers can be passed throughout familial generations without any history of the disease. Educating parents is essential to obtaining an early diagnosis for children affected by Tyrosinemia Type I. Some states screen for this disease using a less reliable method than others. This causes some cases to go undiagnosed. The more parents know about this disease, the more likely they are to piece together the signs and symptoms. They should be aware of the screening procedures, signs and symptoms, further diagnostic tests, treatments available, and current research. The more people who are aware of this disease, the closer to finding a cure we will be. Financial and further research relationships can be made if parents are more knowledgeable about Tyrosinemia Type I. If gene therapy is successful, this will also make breakthroughs for curing other types of genetic disorders such as Cystic Fibrosis.

A focus group of 3 parents with children less than 5 years old born in Alabama or Tennessee was assembled at the University of Alabama in Huntsville. They were given an educational pamphlet regarding Tyrosinemia Type I to review. They also watched a PowerPoint presentation with voice overlay. A discussion was followed about what parents expect from educational materials, which intervention they preferred, anything that should have been included, and general comments or concerns. Data gathered will provide nurses with the best materials to teach parents about Tyrosinemia Type I or other genetic diseases. In this focus group, the PowerPoint presentation was unanimously preferred over the pamphlet, but a more interactive website was identified as a better method than the PowerPoint. The feedback from this focus group will be used to create such a website to better educate parents.
Acknowledgments

Sincere gratitude is given to Elizabeth Barnby, DNP, ACNP-BC, CCRN. This study would not have been possible without her guidance and mentorship. Appreciation is also granted to Ellise Adams PhD, CNM, the supervising faculty of this study. She went beyond the call of duty to ensure this study’s success.
**Introduction**

Tyrosinemia Type I (TT1) is a congenital autosomal recessive metabolic disorder. Autosomal recessive disorders require both parents mutated gene be passed along to the child.

According to Nyhan, Barshop, and Ozand (as cited in Barnby, 2014), TT1 causes a deficiency in fumarylacetoacetate hydrolase (FAH) due to a mutation on chromosome 15q23-25. A genetic mutation is any discrepancy in the gene sequence that causes abnormalities in body functions. FAH, produced by the liver, is part of catabolic pathway that breaks down tyrosine, an essential amino acid. Deficiency in FAH leads to a toxic buildup of several enzymes that will cause neurologic crisis, renal dysfunction, vitamin D-resistant rickets, and end stage liver failure. This disease is often mistaken for Sudden Infant Death Syndrome. If not treated, this disease is fatal before the age of two. According to the Centers of Disease Control, 5-6% of Sudden Infant Death Syndrome fatalities are attributed to metabolic disorders, such as TT1 (Barnby, 2014).

The prevalence of TT1 is 1 or 2 newborns out of every 100,000 births. Many patients need liver transplants which will produce FAH, but this is not a cure. There is medication treatment for TT1, and when diagnosed during new born screening the survival rate is 90% (Barnby, 2014).

New born screening for this disease is done during the neonatal heel stick blood spot test. The heel stick tests for many genetic disorders, and 43 states in the United States test for TT1. It is imperative for pediatric nurses to know what their state tests for in order to educate expecting parents on diseases such as TT1. If their state does not test for TT1, nurses should know the importance of getting a blood screen and where they can obtain one. The only chance of survival for TT1 is diagnosing it during the neonatal stages of life. Symptoms vary in degree from case to case; making TT1 difficult to diagnose if not recognized in the initial screening for the disease. Educating parents about TT1 will help diagnose cases earlier by knowing what to look
for regarding family history and signs and symptoms of the disease. Raising awareness of Tyrosinemia Type I among parents will decrease the rate of children who die from this disease. Educating parents will also increase the number of perspective donors and researchers to further the study of gene therapy to cure the disease.

Many parents are provided written literature at the Pediatrician’s office to learn about specific diseases. With today’s technological advances and ease of access to the public, educational materials should be more interactive to accelerate learning of complicated diseases to the average person. This study sought the opinion of parents with young children in a small focus group setting regarding preferred teaching methods about TT1.

**Literature Review**

Many studies have been conducted on the importance of nursing practice in genetics. Barnoy, Levy, and Bar-Tal (2010) researched how recommendations from nurses and physicians influence decisions to undergo genetic testing. They used a 2 x 2 x 2 factorial design to test subjects. The subjects were interviewed by either an expert or novice nurse, an expert or novice physician, and whether or not a recommendation was actually made. They concluded that it did not matter whether the recommendation came from a nurse or physician, but that they had expert knowledge in the genomics. Genomics is the study of the organization and function of the set of genes. This study demonstrates that knowledgeable nurses are primary educators for genetic screening of TT1. Nurses are also one of the main sources to educate on the pathology of TT1, future testing, treatments, and support groups available.

Benjamin, Birch, Bradley, and Mannion (2013) outlined the “four R’s” that nurses must uphold in regards to genetic health care:

- **Recognize possible genetic conditions and family histories of concern**
• Appreciate the nurses’ Role

• Understand the nurses’ Responsibilities

• Refer appropriately (Benjamin, et al., 2013)

This review identified a need for continuing education for Registered Nurses (RNs) in genomics. Many RNs go on to become certified genetic counselors. According to the Nursing and Midwifery Council (Benjamin, et al., 2013) nurses who graduated before 1995 did not have genomics as part of their curriculum in Bachelor of Science in Nursing studies. This article determined patients would prefer nurses be a primary resource for care and support of families with genetic disorders. This reinforces that nurses should be prepared to be the primary educator for families.

Skirton and Jackson (2013) reviewed different families’ experiences with genetic testing and the nurses’ role. They concluded that the anxiety of testing for disabling diseases keeps many families from pursuing genetic screenings. They examined the importance of a nurses’ knowledge in the genetic testing process and informed consent. The nurse must provide information in an unbiased fashion, know the proper resources to provide, support, and advocate for the families’ decision. This study identifies the need for ethics in regards to informed consent and the importance of respecting families’ wishes with genetic testing.

Kirk and Marshallsay (2013) pinpointed the specific needs families have when children are diagnosed with genetic mutations. They studied 34 articles on parents’ experiences of the diagnosis process and managing children with genetic diseases. The most important aspects that the family needs to understand about their child’s condition are listed below.

• Accept the child’s condition

• Manage the condition on a day-to-day basis
• Meet the normal developmental needs of the child and other family members
• Cope with continuous stress and periodic crises
• Establish a support system. (Kirk and Marshallsay, 2013)

Not only are nurses a priority educator in genetic testing, but they should also know the information and resources families need to cope with the disease. Quality of care goes beyond diagnosing the disease through genetic testing, but should encompass a care plan for the entire family.

This review of literature demonstrates that nurses are the forefront in patient education on genomics and assisting families in care plans for helping children thrive with their disease. TT1 can be screened through carrier testing and newborn heel sticks. When carriers or children are diagnosed early there is a good prognosis for a healthy life. The question that remains is what are the best nursing interventions to educate parents about Tyrosinemia Type I?

Methodology

Purpose of Study

The purpose of this study was to determine which educational material, between a pamphlet and PowerPoint presentation with voice overlay, would best meet the needs of new parents regarding Tyrosinemia Type I. The hypothesis guiding this study was, a PowerPoint presentation with voice overlay will be the best teaching method, as compared to the pamphlet, to educate prospective parents about Tyrosinemia Type I, the screening process, treatments, and current genetic research.

Description of Subjects

The target population for this study was parents, age 19 to 45 years old. Parents of any sex, ethnicity, and health status were recruited for the study. Participants must be a parent of a
child 5 years old or younger born in either Alabama or Tennessee. Both of these states screen for Tyrosinemia Type I by using a more reliable test than other states use. The target population was 4-12 participants. A letter of consent (see appendix A) was signed by the director of the University of Alabama in Huntsville daycare that allowed invitations (see appendix B) to be distributed to parents and placed in backpacks by the daycare staff. Further participant contacts were made through the technique of snowballing.

On the day of the focus group, three subjects meeting the inclusion criteria were in attendance. Although the target population was not acquired, it was viewed more important to obtain data from the present subjects than to cancel the session. This places a limit on the variance of opinion and allows subjects to be more prone to agree with one another than to differ.

**Description of Procedure and Instrumentation**

Elizabeth Barnby, DNP, ACNP-BC, CCRN is an expert on TT1. She published a continuing education credit for RNs on TT1, and is currently performing genetic research to find a cure for TT1 at UAH. The student researcher collaborated with Dr. Barnby to create a PowerPoint presentation with voice overlay and an educational pamphlet. Rebecca Davis, MSN, RN teaches undergraduate courses at UAH College of Nursing. Rebecca Davis reviewed the PowerPoint and pamphlet as an educational expert. After her review, corrections were made to make the material more easily understood. The following documents were sent to the Institutional Review Board: IRB form I, IRB form II, daycare letter, focus group invitation, focus group interview guide, participation consent form, the PowerPoint presentation, and the pamphlet. IRB approval was granted for this study on March 13, 2015 (see appendix C).

The student researcher and faculty adviser were in attendance at the focus group on April 11, 2015 at 12:30 pm at the University of Alabama in Huntsville. Upon arrival, subjects reviewed
and signed a consent form to participate and be audio recorded (see Appendix D). Subjects were encouraged to not use personal identifying information during discussions. Subjects were served lunch as they reviewed an educational pamphlet (see Appendix E) and watched a PowerPoint presentation with voice overlay (see Appendix F). An interview guide (see Appendix G) was used to facilitate group discussion once the educational materials were viewed. Screening methods for TT1 were explained to parents in an effort to inform participants about the screening that their child had in an effort to decrease any fears that their children might have this disease. Specific questions were asked regarding each educational material, what the subjects liked and dislike about it, anything they would change, and how they would like to be presented such materials.

Results

The focus group was conducted in an open discussion format. An interview guide was used to facilitate specific questions regarding the educational materials. The audio recording taken during the focus group was transcribed into text. This transcription was used in content analysis of the narrative data acquired from the participants. Below is a summary of the findings:

Question 1: Is there a preferred method to learning about your child’s health?

It was unanimous among the participants that they would prefer an interactive website to learn more about their children’s health. One participant recommended a gross human anatomy animation that showed all the major organs. A parent could scroll over each organ to learn about the function of the organ and list of juvenile diseases related to that organ. From that disease list a link could take you to another page that explained the disease more in depth. All subjects agreed upon this suggestion and replied they would be more likely to use a similar device as opposed to “relying on myself to read all the papers thrown at you at the hospital”.

Evaluating Parents on TT1

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When asked about the proper timing to teach parents about TT1, initially they unanimously said before giving birth. The consensus was that there is too much information given during the time of birth to focus on learning about rare diseases. One participant mentioned the fact of worrying a parent unnecessarily that their unborn child might have this disease if education were done before birth. This comment swayed the other participant’s opinion to that same conclusion. One participant suggested teaching after the results of the heel stick screen to ease fear and teach to look for particular symptoms in case the screen was unsuccessful at detecting TT1. This swayed the other participants to agree with that scenario. They all concluded that the proper time to educate parents about TT1 would be after the heel stick screen.

Question 2: What did you like about the pamphlet? What did you not like about the pamphlet? Was there anything that should have been included?

The participants agreed that it was difficult to read in its entirety. One claimed “I had to read it twice just to get the gist of it”. They liked the bulleted points the best, and requested to have more bulleted points. “If you can’t follow along in paragraphed text you will not continue to read it, but if you can read short bullets you can skip over the ones you don’t understand and continue to browse the rest.” More pictures were recommended, especially of the liver and kidneys. A diagram explaining what these organ did would be helpful. An image of a damaged liver and organ might put things into perspective.

Question 3: What did you like about the PowerPoint? What did you not like about the power point? Was there anything that should have been included?

The voice overlay was well received and complimented the PowerPoint presentation. It was easier to comprehend the material when someone explained it instead of reading about it for
Educating Parents on TT1

the first time. More pictures of specifically where the liver is, what it does, and an example of it being damaged were requested to be added to the PowerPoint presentation. “If you could virtually show the liver being filled with the succinylacetone, it would help the average person who has no prior medical knowledge have an image of how damaging it is”. It was suggested to add success stories to instill hope for such a dismal disease. “If my child had this I would be beyond thinking straight, especially if I thought that there is no hope”.

Question 4: Did you prefer one over the other and why?

It was unanimous that the PowerPoint presentation was preferred over the pamphlet. They were able to grasp the information better when it was verbally explained, as opposed to reading words that are not easily understood.

Question 5: What suggestions do you have to present the information in a different way that would be more beneficial for parents and improve understanding?

All participants agreed on a website that parents could view at their own leisure. As discussed before, they would like a website that had a general gross anatomy of the organs, what they do, and the different inherited diseases that can be related to them. From this diagram they could be directed to links that further explained the disease process and treatments. When asked if they would want to know what type of research is being done they responded with having the option available if someone wanted to access it. There is so much overwhelming information that links should be provided to further information if wanted. Links to research, genetic counselors, treatment options and support groups should be available, but not discussed on the main page.

Question 6: What do you know now that you did not know before you came today?
The participants said “everything”. All of the information was novel material that the participants were no aware of.

Question 7: Any other concerns, comments, questions, or suggestions?

It was suggested that education material about TT1 should emphasize the signs and symptoms to look for, especially for the states that don’t test for the more reliable screening measure. Areas where it is more prevalent, such as Quebec and France, should request to screen for the more reliable measure if not already done so. They should also have a more in depth class about this disease, such as a segment in a birthing class that would show a presentation similar to this PowerPoint.

The participants were asked if their healthcare providers asked about any history of sudden infant death syndrome (SIDS). They were all warned about it and taught proper sleeping tactics, but none were asked about a history of it in their family. They were also asked what information their healthcare providers gave them about the heel stick test. Two of the participants remember receiving a list of the diseases tested for. They were ensured if the cystic fibrosis screen was positive they would be immediately notified. One participant doesn’t even remember receiving a list, and was just told that they would run some tests.

**Conclusion**

Educating parents is a crucial step to diagnosing more cases of TT1 and bringing recognition to research to help find a cure. Today’s technological advances allows education to be more interactive than just reading about something for the first time. A focus group of parents was used to determine the best ways to educate new parents about TT1. According to the data gathered, an interactive website would be the preferred instrument to teach parents about this disease.
The website should have a gross anatomy diagram of all the vital organs. When the mouse icon is scrolled over an organ the function and list of juvenile diseases associated with it will pop up. Each disease can be selected and create a new link that would explain screening process, history of family, pathophysiology, signs and symptoms, diagnosing, and available treatments. The option to inquire about genetic counseling, specific diets, and current research should be available. Another diagram should show the damage it would cause to that organ or any sub sequential organs involved. Colleges of Nursing could collaborate with the Computer Science Departments to create such a website. This website should be suggested by healthcare providers, including nurses after the newborn heel stick results have returned. The timing of suggesting this education technique would prevent unnecessary fear in expecting parents.

**Nursing Implications and Future Research**

Nurses are viewed as primary patient educators. In order to properly educate patients and their families, nurses should use educational materials that can facilitate learning. Technological advances have created easier ways to instruct parents regarding complicated diseases, such as TT1. The feedback provided during this focus group gives nurses a better idea of what parents expect during teaching and what diagrams would best facilitate learning.

This study was conducted to determine what parents expect from educational sources. A content expert review of researchers, geneticists, and pediatric healthcare providers would be beneficial to gain knowledge about who should provide this education and to determine the appropriate content. Once the website is created, a pre/post test study would determine how well parents were able to grasp the knowledge regarding TT1 on the interactive website.
References


Appendix A: Day Approval

2/5/15

Thank you for agreeing to participate in my honors thesis study entitled, Best nursing interventions for educating parents on Tyrosinemia Type I. An example of the invitation to be given to parents is attached to this notice. Once this study has been approved by UAH Institutional Review Board, more invitations will be provided. Between that date and 3/1/15 you have agreed to give parents these notices by hand or in the backpacks or bags of the children who attend your facility.

Please know that your assistance with this work is greatly appreciated. If you have further questions, please contact me or my research adviser using the information below.

Julianna Mize
(334)332-0431
Jm0081@uah.edu

Ellise D. Adams PhD, CNM
Associate Professor, Nursing
256-824-2442
Ellise.adams@uah.edu

UAH University Center Child Care
Director signature

Feb 5, 2015
Appendix B: Focus Group Invitation

YOU ARE INVITED TO A
Luncheon

Please join us during the month of March at the University of Alabama in Huntsville. Specific date to be communicated upon receipt of interest.

You have been invited to share your opinion and contribute to educational techniques appropriate for parents with children under the age of 12. Lunch is free! Human nursing student Jovena Knox will be facilitating this focus group about neonatal diseases detected by the heel stick test.

Please email jknox@bch.edu for more information.
Appendix C: IRB Approval

Julianna Mize and Ellise Adams  
Colleg of Nursing  
March 13, 2015

Dear Julianna Mize and Dr. Adams,

The UAH Institutional Review Board of Human Subjects Committee has reviewed your proposal, *Best Nursing Interventions for Educating Parents on Tyrosinemia Type I*, and found it meets the necessary criteria approval. Your proposal seems to be in compliance with this institution's Federal Wide Assurance (FWA) 00019998 and the DHHS Regulations for the Protection of Human Subjects (45 CFR 46) and has been classified as expedited.

Please note that this approval is good for one year from the date on this letter. If data collection continues past this period, you are responsible for processing a renewal application a minimum of 60 days prior to the expiration date.

No changes are to be made to the approved protocol without prior review and approval from the UAH IRB. All changes (e.g., a change in procedure, number of subjects, personnel, study locations, new recruitment materials, study instruments, etc.) must be prospectively reviewed and approved by the IRB before they are implemented. You should report any unanticipated problems involving risks to the participants or others to the IRB Chair.

If you have any questions regarding the IRB’s decision, please contact me.

Sincerely,

Pam O’Neal PhD, RN  
IRB Chair  
Associate Professor  
College of Nursing, University of Alabama in Huntsville,  
207 Nursing Building, Huntsville, AL 35899  
phone: 256.824.5191 or 6100 and fax: 256.824.2850 email: irb@uah.edu
Appendix D: Consent Form

Consent Form

Educating parents on Tyrosinemia Type I

You are invited to participate in a focus group about education techniques appropriate for parents with children under the age of 12 months related to newborn diseases. This study is designed to help us to better understand how nurses can educate parents on complex diseases, screening, treatments, and genetic research. The student researcher is Julianna Mize, from the Nursing College of the University of Alabama in Huntsville. She may be contacted by email: Jm0081@uah.edu. Supervising faculty: Dr. Ellise Adams may be contacted through email: Ellise.Adams@uah.edu.

PROCEDURE TO BE FOLLOWED IN THE STUDY: Once written consent is given, you will join a focus group with 4-12 other parents of children less than 12 months old. The student researcher and faculty advisor will be in attendance. You will each be given a pamphlet to review and watch a PowerPoint presentation. You will then discuss it as a group. This session will last 2 hours and lunch will be provided. The entire session will be audiotaped. Please do not use any identifying information during the focus group.

DISCOMFORTS AND RISKS FROM PARTICIPATING IN THIS STUDY: There are no unforeseen physical, psychological, social, economic, and/or legal risks involved with this study. Alabama and Tennessee both screen for this disease using the most accurate test. There should be no added anxiety that your child has this disease, because you would have been notified after the hee1 stick test results were received. By signing this form you understand that this session will be recorded and contents will be transcribed to determine results of the study. You will be requested not to use your name, and all names shall be omitted during transcription if applicable.

EXPECTED BENEFITS: Benefits include education on Tyrosinemia Type I and lunch provided.

CONFIDENTIALITY OF RESULTS: Participant numbers will be used to record your data, and these numbers will be made available only to those researchers directly involved with this study, thereby ensuring strict confidentiality. This consent form will be destroyed within 12 months. The data from your session will only be released to those individuals who are directly involved in the research and only using your participant number.

FREEDOM TO WITHDRAW: You are free to withdraw from the study at any time. You will not be penalized because of withdrawal in any form. Investigators reserve the right to remove any participant from the session without regard to the participant’s consent.

CONTACT INFORMATION: If any questions should arise about this study or your rights as a participant, you may contact the student researcher at any point in the research process. You may contact Julianna Mize in UAH College of Nursing at Jm0081@uah.edu. If you have questions about your rights as a research participant, or concerns or complaints about the research, you may contact the Office of the IRB (IRB) at 256.824.6101 or email Dr. Pam O’Neal at irb@uah.edu.

If you agree to participate in our research please sign and date below.
This study was approved by the Institutional Review Board at UAH and will expire in one year from <date of IRB approval>.

Name (Please Print)  Signature  Date

04/2013
Thank you for participating in this study!

The University of Alabama in Huntsville College of Nursing has teamed up with the College of Biology to research Tyrosinemia Type I (TT1) and the effects of the medication to treat this disease on mental function. Currently research is being done at UAH to examine how the present therapy alters mental function in mice with the disease. Additionally, researchers are studying ways to correct the mutation so that children with TT1 can lead a normal life. These research efforts could also lead to cures for other genetic disorders such as Cystic Fibrosis.

References


Appendix E: Educational Pamphlet (Inside)

Tyrosinemia Type I

What is it?
Tyrosinemia Type I (TTI) is an inherited genetic disorder that is fatal if not caught early. The liver is unable to produce a substance that breaks down the Amino Acid, Tyrosine. There is about 1 case for every 100,000 births. It is more common in areas of Quebec, Canada. If not treated, death can occur before the age of two. These deaths are often labeled Sudden Infant Death Syndrome (SIDS).

How can my child get this?
TTI is an autosomal recessive genetic disease. Both parents must be carriers of the disease to pass it on to their children. If both parents are carriers, the child has a 25% chance of inheriting the disease, a 50% chance of becoming a carrier, and a 25% chance of not being affected. Genetic counselors can advise families about risks and tests to identify the disease. Carriers can continue to pass on the TTI gene through generations without symptoms.

How will I know if my child has TTI?
All states currently perform screening for TTI as part of the newborn screening heel stick. Unfortunately, some states screen for the measurement of tyrosine, which can make false negatives. Testing for levels of the toxic substance succinylacetone that builds up in the body as a result of this disease is a more reliable screening method. There is a study currently being done at UAH to determine which states test for the more reliable marker. If there is a family history of TTI and your state does not test for the more reliable marker, Succinylacetone, then a more in depth screening process should be requested.

Alabama and Tennessee both screen for succinylacetone, which is the most reliable screening tool for TTI. Parents should not be alarmed that their child could have TTI if they have not been contacted following the heel stick.

If the child tests positive for succinylacetone during this initial screen, then further tests will be performed to confirm the diagnosis of TTI.

Signs and Symptoms
- Vary according to degree of illness.
- Nausea/vomiting, especially after foods high in protein
- A generalized yellow/orange appearance (Jaundice)
- “Burnt cabbage” smelling urine
- Slow growth
- Bloated abdomen
- Low blood sugar
- Painful neurologic crisis

How is TTI treated?
There is no cure for TTI although treatments are available to manage it. Orfadin (Nitisone) is the only medication used for TTI. This medication must also be coupled with a low protein diet. The earlier this disease is treated, the better the chances of survival. Even with treatment, many patients will need liver and/or kidney transplants.

THERE IS HOPE!
Scientists at UAH are hard at work trying to understand and develop treatment. Gene therapy is being studied to hopefully find a cure.
Appendix F: PowerPoint Presentation (Slides 1-4)

Thank You for Participating!

Your participation is greatly appreciated in assisting us to find better ways of educating parents about their children’s health. Alabama and Tennessee both screen for Tyrosinemia Type I using a highly reliable test during the heel stick screen. It is highly unlikely that your child would have this disease without this screening measure detecting it. This study is being performed to help raise awareness of the disease, especially for parents who live in other areas which do not use this extremely accurate screening method.

What is Tyrosinemia Type I?

- Tyrosinemia Type I (TTI) is an inherited Genetic Disorder.
- The liver is unable to produce a substance that breaks down Amino Acid Tyrosine
- It is fatal if not diagnosed and treated early

How common is it?

- There is about 1 diagnosed case for every 100,000 births
- It is more common in certain areas, such as Quebec, Canada.
- These deaths are often labeled as Sudden Infant Death Syndrome.
Appendix F: PowerPoint Presentation (Slides 5-8)

Is my child at risk for TTI?

- TTI is a metabolic autosomal recessive disease

It’s a WHAT?!

- Autosomal recessive means that both parents must at least be carriers of the disease for the child to have a chance of inheriting TTI.
- If both parents are carriers then:
  - There is a 25% chance the child will have TTI
  - 50% chance that the child will just be a carrier with no symptoms
  - 25% chance the child will not be affected

Just like the punnet square from biology!

How will I know if my child has TTI?

- All states currently screen for TTI as part of the newborn heel stick.
- Some states use a less reliable marker due to cost of testing.
- There is currently a research project being conducted at UAH college of nursing to determine which states screen for the more reliable marker.
Educating Parents on TT1

Appendix F: PowerPoint Presentation (Slides 9-12)

Symptoms of TTI
- Nausea and vomiting
- A yellow/orange appearance
- Burnt cabbage smelling urine
- Slow growth
- Inability to gain weight
- Low blood sugar
- Painful Neurogenic crisis

How is TTI treated?
- There is no known cure for TTI
- There is only one drug to treat it: Orfadin (Nitisnone).
- The drug must be coupled with a low protein diet.
- Even with treatment, liver and often kidney transplants are needed.

THERE IS HOPE!
The University of Alabama
In Huntsville College of Nursing
and College of Biology are
Researching the use of gene therapy to help find a cure. If successful, this could also make more breakthroughs to cure other genetic diseases such as cystic fibrosis.

References
Appendix G: Focus Group Interview Guide

Thank you so much for participating in this focus group. You input will greatly help us to create better educational materials to parents with children who have genetic diseases. My name is Julianna Mize, and I am a honors student in UAH BSN program. Dr. Ellise Adams is my faculty adviser and an instructor for the College of Nursing. She will be assisting today during this focus group.

Educating patients and families is an important role of nursing care. Typically the educational material provided by healthcare professionals comes in the form of a pamphlet. We are trying to determine a way to provide educational material that might be more beneficial to parents of children with Tyrosinemia Type I. TTI is a genetic disease that requires major life changes to control symptoms. Proper education on this disease will be the first step to living with TTI. This focus group will help us develop an educational guide to these parents.

You will receive a pamphlet to review and then watch a power point presentation. We will then have an open discussion about ways to improve these educational materials. I welcome all input, including criticism or differing points of view. This session will be audio recorded. For the sake of your confidentiality, please do not use any identifying information during this focus group. Please enjoy lunch!

Is there a preferred method to learning about your child’s health?

What did you like about the pamphlet? What did you not like about the pamphlet? Was there anything that should have been included?

What did you like about the power point? What did you not like about the power point? Was there anything that should have been included?

Did you prefer one over the other and why?

What suggestions do you have to present the information in a different way that would be more beneficial for parents and improve understanding?

What do you know now that you did not know before you came today?

Any other concerns, comments, questions, or suggestions?