Tyrosinemia Type 1: An Overview of Nursing Care

Elizabeth Barnby

Tyrosinemia type 1 (TT1) is an inherited metabolic disease that can be fatal when not detected early by newborn screening. In the past, children with TT1 had a poor prognosis due to organ failure and neurologic crisis during infancy. Recent improvements in newborn screening have changed the prognosis of affected children. Measurement of succinylacetone by tandem mass spectrometry provides early identification and the opportunity to manage TT1 as a chronic disease. Treatment includes genetic counseling, dietary management, pharmacotherapy, metabolic crisis prevention, and whole organ transplant. Nursing care is critical to successful management when it is based on a clear understanding of the pathophysiology. This overview of nursing care will provide specific recommendations to reduce complications and enhance the quality of life for children with TT1.

The deficiency is caused by a mutation on chromosome 15q23-25 (Nyhan, Barshop, & Ozand, 2005; Paradis, 1996). The disease is an autosomal recessive disorder that is expressed when a child receives the trait or mutation from both parents. In the carrier state, the mutation is harmless but can be passed on to offspring.

An interruption in the tyrosine catabolic pathway causes a metabolic gridlock in the human body with toxic substances accumulating in tissues throughout the body. Treatment can prevent the accumulation of these toxic substances. The combination of improvements in newborn screening and pharmacotherapy has improved outcomes (Nobili et al., 2010).

The FAH enzyme at the terminal end of the tyrosine catabolic pathway is crucial to the breakdown of tyrosine. When it is absent, toxic metabolites higher up the pathway accumulate and cause disease (Scott, King, & Trahms, 2008). Occasionally, a spontaneous mutation can occur that produces pockets of normal enzyme activity, and this produces variable phenotypic expression (Nakamura, Tanaka, Mitsubuchi, & Endo, 2007). Because phenotypic expression varies, two children with the exact same chromosomal mutation can have varying degrees of disease severity.

TT1 (McHugh et al., 2011; Schunemann et al., 2008). When TT1 is identified by enhanced newborn screening methods, the disease is treatable (Turgeon et al., 2008). If not identified on newborn screening, children with the disease can present critically ill in metabolic acidosis. Children with inborn errors of metabolism are identified post-mortem when newborn screening fails to identify the disease in the neonate (Bennett & Rinaldo, 2001; CDC, 2003; Chace et al., 2001). Early identification and diagnosis are life-saving for these metabolically unstable patients. The pediatric nurse needs to understand the pathophysiology of the disease and nursing interventions that will improve patient outcomes to successfully intervene. An overview of the pathophysiology and nursing care will enhance outcomes when applied at the bedside.

Pathophysiology

TT1 is characterized by the child’s inability to break down tyrosine. Tyrosine is an essential amino acid contained in protein that the body needs to perform cellular functions. Normally, the enzyme fumarylacetoacetate hydrolase (FAH) catalyzes tyrosine, but children with TT1 have a deficiency of this essential enzyme.

Acknowledgment: This work would not have been possible without the wise guidance of Dr. Karen Frith.

Statements of Disclosure: Please see page 67 for statements of disclosure.
Tyrosinemia Type 1: An Overview of Nursing Care

FAH deficiency leads to an accumulation of fumarylacetoacetate, maleylacetoacetate, succinylacetoacetate, and succinylacetone. Maleylacetoacetate causes renal tubular dysfunction in a Fanconi-like syndrome of renal failure and vitamin D-resistant rickets (Jacobs, van Beurden, Klomp, Berger, & van den Berg, 2006). Fumarylacetoacetate and maleylacetoacetate cause hepatocyte injury that can result in end stage liver disease, bleeding, and hepatocellular carcinoma (Orejuela, Jorquera, Bergeron, Finegold, & Tanguay, 2008). Succinylacetone is an inhibitor of the heme synthesis pathway, causing neurotoxicity that is similar to lead poisoning or aminolevulinic acid dehydratase (ALAD) deficiency porphyria (Wyllie & Hyams, 2006). The neurotoxicity can cause muscle paralysis and respiratory arrest (Krous, 2010; Turgeon et al., 2008).

Diagnosis

Newborn screening for TT1 is performed by tandem mass spectrometry. Tandem mass spectrometry is an accurate way to measure newborn blood spot specimens for the presence of disease markers. Each state mandates a different newborn screening panel for their population of newborns (Howell, 2009). Most states are now using the improved newborn screening test for TT1 that measures succinylacetone (Morrisey, Sunny, Fahim, Lubowski, & Caggana, 2011). Measurement of tyrosine is neither specific nor sensitive for the identification of TT1 (Allard, Grenier, Korson, & Zytkovicz, 2004). Succinylacetone measurement by tandem mass spectrometry is accurate and reliable for diagnosing TT1 in the newborn (Turgeon et al., 2008). Early identification of this life-threatening disease provides an opportunity to intervene prior to symptom onset.

The clinical presentation is complicated. Symptoms can be severe or chronic depending on phenotypic expression of the disease (Nakamura et al., 2007). Some children present with failure to thrive and poor growth, but others have an acute onset of metabolic acidosis, hepatomegaly, ascites, coagulopathy, and neurologic crisis (Croffie, Gupta, Chong, & Fitzgerald, 2010). Metabolic crisis is precipitated by hypoglycemia.

Treatment

Children with TT1 must maintain strict metabolic control of their disease. Parents play a key role in metabolic control by maintaining dietary restriction of protein consumption and avoidance of protein catabolism (Ashorn, Pitkanen, Salo, & Heikinheimo, 2006). Typical treatment requires intensive developmentally appropriate patient and family education. An age-appropriate guide is useful to address these educational needs (see Table 1).

Avoiding protein catabolism is difficult in the presence of an acute viral illness. When unable to eat, the body naturally breaks down protein stores to maintain homeostasis. The breakdown of protein causes the level of amino acid tyrosine to rise and results in a metabolic crisis (Claudius, Fluharty, & Boles, 2005).

During fasting, the body produces ketones. Urine ketones can be used as a metabolic indicator of protein catabolism. Ketones are a breakdown product of fatty acid metabolism. Ketones in the urine should be managed as an emergency. When ketotic, it is essential for the child to consume glucose to prevent protein catabolism (Claudius et al., 2005). If the child is unable, intravenous therapy can prevent complications associated with increased tyrosine levels. An antiemetic may be ordered if nausea and vomiting are present. Antiemetics can also be prescribed as needed to prevent future episodes of ketosis and adverse outcomes (Fedorowicz, Jagannath, & Carter, 2011).

Without the steady consumption of fat and carbohydrates, muscle protein is metabolized and tyrosine levels rise. As tyrosine levels increase, toxic metabolites build up. One toxic metabolite is succinylacetone, which causes ALAD porphyria in a complex biochemical process that interferes with heme metabolism. This biochemical interaction results in neurologic crisis (Anderson et al., 2005).

Neurologic Crisis

Neurologic crisis can present as a syndrome of severe pain, paralysis, and respiratory arrest. Neurologic effects of FAH deficiency in TT1 include severe abdominal pain, extremity pain, nausea, vomiting, weakness, and altered level of consciousness. Succinylacetone inhibits conversion of Delta-aminolevulinic acid to porphobilinogen in the heme synthesis pathway. Delta-aminolevulinic acid is neurotoxic. The symptoms are similar to the symptoms of porphyria. Lead is also an inhibitor of the conversion of Delta-aminolevulinic acid. Inhibition of the pathway causes neuropathy, neurologic crisis, paralysis, and respiratory distress (Mitchell et al., 1990). Mental retardation and seizure disorders have also been reported in children with errors in the metabolism of tyrosine (Palmer, 2006; Rocha et al., 2000). With careful medical management and nursing care, this complication is preventable. The ability to prevent neurologic deficits offers financial and ethical incentive to maintain high quality newborn screening for the disease (Bailey, Skinner, & Warren, 2005).

Neurologic crisis is preventable if the family is educated and health care providers respond appropriately to the metabolic crisis. The most important way to prevent neurologic crisis is prompt administration of IV glucose solutions, usually 10% or higher depending on severity of symptoms and serum glucose measurement. Arterial blood gas measurement with calculation of anion gap can indicate severity of metabolic acidosis. A written plan is advised for the family to have with them in the event of an emergency. The written emergency plan can advise providers of treatment plans and how to contact medical genetics experts for consultation. The American College of Emergency Physicians (ACEP) and the American Academy of Pediatrics (AAP) provide an online template that is useful in developing this document (AAP, 2010), which can be accessed online at http://www.aap.org/advocacy/blark form.pdf (ACEP, 2008).

Diet and Pharmacotherapy

TT1 is a treatable disease. An experienced team that includes a dietician, medical geneticist, advanced practice nurse, psychologist, school nurse liaison, and other practitioners will need to collaborate as an interdisciplinary team to enhance outcomes (Paradis, 1996). The treatment includes a low-protein diet with medical food or formula, as well as the medication nitisinone (Orfadin®). The orphan drug nitisinone is used to treat TT1. Nitisinone shifts the meta-
Table 1.
Age-Appropriate Care and Educational Needs of Children with TT1 and Families

<table>
<thead>
<tr>
<th>Age of Patient</th>
<th>Developmentally Appropriate Disease Management Timeline for Nursing Education of Caregivers and Patient</th>
</tr>
</thead>
<tbody>
<tr>
<td>0 to 6 months</td>
<td>*This is merely a guide that should be adjusted according to parent-child shared management preferences.</td>
</tr>
<tr>
<td></td>
<td>- Caregivers learn to prepare prescribed formula using chemistry scale.</td>
</tr>
<tr>
<td></td>
<td>- Caregivers learn to complete 24-hour diet history for review with dietician and medical geneticist.</td>
</tr>
<tr>
<td></td>
<td>- Caregivers learn disease management and pathophysiology of protein metabolism in patient with TT1.</td>
</tr>
<tr>
<td></td>
<td>- Caregivers learn of financial resources to purchase medical food (varies by state and insurance).</td>
</tr>
<tr>
<td></td>
<td>- Caregivers learn importance of not showing signs of aversion to medical food since feeding is easily influenced by social situations.</td>
</tr>
<tr>
<td></td>
<td>- Medical food is life-sustaining and must be a positive social experience to promote future healthy dietary habits.</td>
</tr>
<tr>
<td></td>
<td>- Caregivers learn to schedule appointments with dietician every three months.</td>
</tr>
<tr>
<td></td>
<td>- Caregivers learn to notify health care providers of episodes of ketosis (inability to consume calories can result in protein overdose when protein catabolism occurs).</td>
</tr>
<tr>
<td></td>
<td>- Caregivers verbalize how protein catabolism can cause toxic metabolites to increase and become life-threatening.</td>
</tr>
<tr>
<td></td>
<td>- Caregivers are given written emergency action plan to carry at all times to advise emergency providers of genetic provider contacts and emergency care of a child with TT1.</td>
</tr>
<tr>
<td></td>
<td>- Caregivers learn to schedule yearly eye examinations to rule out tyrosine accumulation in corneas.</td>
</tr>
<tr>
<td></td>
<td>- Caregivers administer Orfadin® as prescribed (Orfadin® must be refrigerated).</td>
</tr>
<tr>
<td>6 months</td>
<td>- Caregivers introduce low-protein foods.</td>
</tr>
<tr>
<td></td>
<td>- Caregivers learn to use books to look up protein content of all foods and enter on diet history record.</td>
</tr>
<tr>
<td></td>
<td>- Caregivers learn to meet exact protein requirements (varies by weight and medical food prescription).</td>
</tr>
<tr>
<td></td>
<td>- Caregivers learn to facilitate consumption of exact protein requirement.</td>
</tr>
<tr>
<td></td>
<td>- Caregivers given low-protein cookbook, and introduced to online sources of low-protein foods and parent support groups.</td>
</tr>
<tr>
<td></td>
<td>- Caregivers learn safe blood levels of tyrosine.</td>
</tr>
<tr>
<td></td>
<td>- Caregivers learn to prevent ketosis.</td>
</tr>
<tr>
<td></td>
<td>- Caregivers learn to measure ketones in urine and act appropriately to reverse ketosis with glucose solution or medical intervention as needed.</td>
</tr>
<tr>
<td>6 to 7 months</td>
<td>- Caregivers introduce cup for low-protein formula as prescribed.</td>
</tr>
<tr>
<td>8 to 9 months</td>
<td>- Caregivers introduce finger food within specific protein restriction.</td>
</tr>
<tr>
<td>10 to 15 months</td>
<td>- Caregivers consider weaning to cup; continue medical food as prescribed by cup.</td>
</tr>
<tr>
<td></td>
<td>- Caregivers learn to prepare low-protein meals.</td>
</tr>
<tr>
<td></td>
<td>- Siblings may assist with low-protein diet preparation if interested.</td>
</tr>
<tr>
<td>2 to 3 years</td>
<td>- Patient learns to distinguish acceptable foods.</td>
</tr>
<tr>
<td></td>
<td>- Patient begins to learn to swallow capsule as tolerated.</td>
</tr>
<tr>
<td>4 to 5 years</td>
<td>- Patient learns to count foods.</td>
</tr>
<tr>
<td></td>
<td>- Patient begins to learn to weigh medical food and other low-protein foods on chemistry scales.</td>
</tr>
<tr>
<td></td>
<td>- Begins to measure grams of medical food and assist with mixing of medical food.</td>
</tr>
<tr>
<td>5 to 6 years</td>
<td>- Patient assists caregivers with formula preparation.</td>
</tr>
<tr>
<td></td>
<td>- Caregivers teach child how to interact with other school-age children and satisfy their curiosity about specific dietary restrictions in a positive way.</td>
</tr>
<tr>
<td></td>
<td>- Caregivers teach educators to monitor diet at school and prevent socially inappropriate behaviors that inhibit healthy diet habits.</td>
</tr>
<tr>
<td>7 to 10 years</td>
<td>- Patient begins to list foods on 14-hour diet recall.</td>
</tr>
<tr>
<td></td>
<td>- Patient learns to pack school lunch with parental assistance.</td>
</tr>
<tr>
<td></td>
<td>- Patient chooses appropriate afterschool snacks.</td>
</tr>
<tr>
<td></td>
<td>- Patient learns to look up protein grams and read labels of foods.</td>
</tr>
<tr>
<td></td>
<td>- Patient learns to request restaurant menus with protein content when available.</td>
</tr>
<tr>
<td>10 to 12 years</td>
<td>- Patient begins to prepare and consume medical food independently with parental monitoring.</td>
</tr>
<tr>
<td></td>
<td>- Patient learns to prepare simple entrees with parental supervision.</td>
</tr>
<tr>
<td></td>
<td>- Patient learns safe blood levels of tyrosine (below 500).</td>
</tr>
<tr>
<td></td>
<td>- Patient learns to plot height and weight during clinic visits.</td>
</tr>
<tr>
<td></td>
<td>- Patient learns why height and weight is important for overall health and development.</td>
</tr>
<tr>
<td></td>
<td>- Patient attends yearly metabolic camp if available through referral in genetics clinic. (Children with TT1 usually can attend metabolic camp with children that have PKU since disease management is similar.)</td>
</tr>
</tbody>
</table>

continued on next page
sources:

**If liver transplant is performed, diet therapy is no longer necessary.

**Tyrosinemia Type 1: An Overview of Nursing Care**

Table 1. (continued)

<table>
<thead>
<tr>
<th>Age of Patient</th>
<th>Developmentally Appropriate Disease Management Timeline for Nursing Education of Caregivers and Patient</th>
</tr>
</thead>
<tbody>
<tr>
<td>13 to 14 years</td>
<td><em>This is merely a guide that should be adjusted according to parent-child shared management preferences.</em></td>
</tr>
<tr>
<td></td>
<td>• Patient learns menu planning.</td>
</tr>
<tr>
<td></td>
<td>• Patient expands on preparation of meals with parental supervision.</td>
</tr>
<tr>
<td></td>
<td>• Patient keeps calendar for blood draws.</td>
</tr>
<tr>
<td></td>
<td>• Patient assumes responsibility for food records.</td>
</tr>
<tr>
<td></td>
<td>• Patient assists with grocery shopping for appropriate low-protein foods.</td>
</tr>
<tr>
<td></td>
<td>• Patient attends low protein cooking classes with peers with similar low-protein diet needs.</td>
</tr>
<tr>
<td></td>
<td>• Patient learns to shop for low-protein foods online and in person if available.</td>
</tr>
<tr>
<td></td>
<td>• Patient learns to interact with health care providers with autonomy.</td>
</tr>
<tr>
<td>15 to 17 years</td>
<td>• Patient assumes responsibility for remembering recent lab results.</td>
</tr>
<tr>
<td></td>
<td>• Patient schedules clinic appointments.</td>
</tr>
<tr>
<td></td>
<td>• Patient schedules yearly eye exams to rule out tyrosine accumulation in corneas.</td>
</tr>
<tr>
<td>18 years</td>
<td>• Patient transition to adult clinic care.</td>
</tr>
<tr>
<td></td>
<td>• Patient manages diet and follow-up care.</td>
</tr>
<tr>
<td></td>
<td>• Patient learns to access assistance when needed.</td>
</tr>
</tbody>
</table>

**Liver Transplant**

Decreased hepatocellular apoptosis in the presence of toxic metabolites in the liver increase the risk for hepatocellular carcinoma (HCC) in children with TT1. Consultation with a pediatric hepatologist and yearly ultrasounds, CT scans with contrast, or gadolinium-enhanced MRI can detect hepatic nodules early (Lauenstein et al., 2007). Alpha-fetoprotein (AFP) levels should be followed as well. Elevations of AFP may indicate HCC. Early referral for liver transplant before nodules are greater than 2 cm can be life-saving (Coelli et al., 2006).

Unfortunately, even with early nitisinone treatment, some children develop HCC and require liver transplant. After liver transplant, the new liver allograft provides FAH enzyme to metabolize dietary tyrosine.
transplant, the low-protein diet is unnecessary. The liver transplant team and pediatric hepatologists manage maintenance of immunosuppression. Liver transplant is considered life-saving, but it is not a cure for TT1. The risk of rejection, infection, and malignancy after liver transplant is significant (Arnon et al., 2011; Heffron et al., 2010).

Liver transplant decreases the tyrosine load, but it does not abolish renal exposure to fumarylacetoacetate (FAA). Low-dose nitisinone can be used after liver transplant if succinylacetone is found in the urine. It is thought that low-dose nitisinone may slow nephropathy and decrease the risk of renal carcinoma from exposure to FAA and succinylacetone in the kidneys. The kidneys also metabolize tyrosine and may require transplant if end stage renal disease develops (Jacobs et al., 2006).

Nursing Care

The care of the child and family with TT1 requires an interdisciplinary, collaborative team approach. Following the tenets of family-centered care, the family is considered an essential part of the team and should be included in care planning in the acute care and outpatient care settings. At different stages of disease progression, treatment decisions will need to be made that enhance both the quantity and quality of life for the child and caregivers. The nurse assumes the role of patient advocate and educator to assure that the family participates in care planning and understands the chronic nature of this lifelong condition. Anticipatory guidance on immunizations, developmental milestones, and future therapy will enhance the quality of life (Vinson, 2002).

Prior to transplant, the primary treatment is prevention of metabolic deterioration and neuro-

---

Table 2.
Medical Food and Formula Resource List

<table>
<thead>
<tr>
<th>Company Name</th>
<th>Contact</th>
<th>Tyrosinemia Formula</th>
<th>Low-Protein Foods</th>
</tr>
</thead>
<tbody>
<tr>
<td>Cambrooke Foods®</td>
<td>508.782.2300 <a href="mailto:orders@cambrookefoods.com">orders@cambrookefoods.com</a></td>
<td><strong>Formula Per prescription and</strong></td>
<td>Breads</td>
</tr>
<tr>
<td></td>
<td></td>
<td><strong>dietician management</strong></td>
<td>Pasta</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Baking ingredient</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Ready Meals</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Cheese</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Rice</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Snacks</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Soups and seasonings</td>
</tr>
<tr>
<td>Mead Johnson™</td>
<td>1.847.832.2420 <a href="http://www.meadjohnson.com">http://www.meadjohnson.com</a></td>
<td>1.812.429.5000</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Abbott Nutrition</td>
<td>1.800.227.5767 <a href="http://Abbottnutrition.com">http://Abbottnutrition.com</a></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Nutricia Advanced Medical Nutrition LoProfin</td>
<td>1.800.365.7354 Myspecialdiet.com 1.877.636.2283</td>
<td>1.800.365.7354 Myspecialdiet.com 1.877.636.2283</td>
<td>Baking mix</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Cereal</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Crackers</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Pasta</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Cake mix</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Chips</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Rice</td>
</tr>
<tr>
<td>Applied Nutrition</td>
<td><a href="http://medicalfood.com">http://medicalfood.com</a> 1.800.605.0410 1.973.734.0023</td>
<td></td>
<td>Snacks</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Candy</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Chips</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Baking mixes</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Cereal</td>
</tr>
<tr>
<td>Dietary Specialties</td>
<td><a href="http://www.dietspec.com">http://www.dietspec.com</a></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Sauces</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Mixes</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Puddings</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Gels</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Bread</td>
</tr>
</tbody>
</table>

---
logic crisis with diet therapy and nitisinone. These therapies should be implemented in a developmentally appropriate way (see Table 2) (Miller & MacDonald, 2006; Taylor, 1996).

*Growth and Development*

Assessment of dietary intake by evaluation of a 24-hour food diary while on a low-protein diet is an important nursing intervention. A three-day dietary history can also provide critical information about the dietary challenges families face. A healthy diet within food restrictions will enhance normal growth and development. Growth measurements at routine visits should include height and weight measurements. Growth should be monitored by comparison to children of comparable age and gender with normalized values on standardized growth charts (Miller & MacDonald, 2006).

Children and families managing the strict diet will need detailed instructions and feedback on progress to be successful. An individualized nutritional assessment by the metabolic team will provide the dietary prescription, and nurses should educate families about specific measures that improve compliance. Careful attention to delivery of medical food in a socially and developmentally appropriate manner can make all the difference when compliance is an issue. The nursing goal is for the child to eventually self-manage the disease (Kieckhefer & Trahms, 2000; Kieckhefer, Trahms, Churchill, & Simpson, 2009). Co-management with family member supervision can be developed as early as two years of age and continue until the patient is able to self-manage his or her disease in adulthood. A table to outline the care and educational needs of the family at each developmental stage can be a useful tool when planning individualized care (see Table 1).

*Patient and Family Education*

Early recognition and management of complications to minimize the negative impact through appropriate care strategies will improve long-term outcomes. Minor viral illnesses can become serious if calories are not consumed. The catabolism of protein stores can induce a metabolic crisis. It is important for families of children with TT1 to be advised of this danger. They should be instructed in how to test the urine for ketones. Ketones in the urine can alert families to the danger.

Acute exacerbations should be managed in an acute care facility with close attention to glucose levels and monitoring for metabolic acidosis caused by a build up of toxic metabolites during fasting. Parents should be taught when to bring the patient to the hospital, when to consult the genetics team, and when to give Pedialyte® or glucose solutions to avoid this type of metabolic crisis (Claudius et al., 2005).

Infection prevention is important. Vaccine-preventable diseases can be avoided by routine vaccination to include yearly flu vaccines. After transplant, no live virus vaccines can be administered (Mack et al., 1996). Teaching families simple infection control, such as hand washing, can be helpful when developmentally appropriate.

*Psychosocial Support*

The complexity of disease management and variety of complications experienced by patients and their families make psychosocial supportive care and patient education essential. The psychosocial support of the family is a critically important role for the nurse. The nurse’s role can minimize the impact of associated complications and delay disease progression. Support of the family and child can facilitate developmental achievement and allow the patient to live a happy and productive life. Green (2009), describes what it is like to manage a chronic disease that results in whole organ transplant. In an ethnographic design, Green (2009) isolated four key themes identified by parents of pediatric heart transplant recipients: coping with life, and constantly responsible, worried, and blessed. Parents of these children are under immense psychological pressure, but often feel blessed by the gift of a second chance at life for their child.

Coffey (2006) identified seven themes that describe how parents feel about parenting a child with a chronic disease. This metasynthesis reveals the complex emotions and hardship these families deal with on a daily basis. The seven themes revealed by this metasynthesis described parental feelings about survival as a family, the constant worry and struggles, the burden of disease and an attempt to take charge, critical periods during the illness, and bridges to the outside describing the isolation felt by some parents. Although parents often feel isolated and anxious, the nurse’s role can provide critical emotional support, sensitivity to their struggle, education on technical care, and effective coping strategies that lighten their burden. Caring is truly the first step in assisting families dealing with the demands of managing a complicated disease such as TT1.

**References**


Tyrosinemia Type 1: An Overview of Nursing Care

Deadline for Submission: April 30, 2016

PED 1403

To Obtain CNE Contact Hours
1. For those wishing to obtain CNE contact hours, you must read the article and complete the evaluation through Pediatric Nursing’s Web site. Complete your evaluation online and your CNE certificate will be mailed to you. Simply go to www.pediatricnursing.net/ce
2. Evaluations must be completed online by the above deadline. Upon completion of the evaluation, a certificate for 1.3 contact hour(s) will be mailed.

Fees – Subscriber: Free Regular: $20

Goal
The purpose of this learning activity is to enable the reader to discuss tyrosinemia type 1 (TT1), its pathophysiology, diagnosis, treatment, and implications for nursing.

Objectives
1. Define tyrosinemia type 1 (TT1).
2. Discuss the pathophysiology of TT1.
3. Explain treatment options for TT1.
4. Discuss nursing implications when treating patients with TT1.

Statement of Disclosure: The author(s) reported no actual or potential conflict of interest in relation to this continuing nursing education activity.

The Pediatric Nursing Editorial Board members reported no actual or potential conflict of interest in relation to this continuing nursing education activity.

This independent study activity is provided by Anthony J. Jannetti, Inc. (AJJ).
Anthony J. Jannetti, Inc. is accredited as a provider of continuing education by the American Nurses Credentialing Center’s Commission on Accreditation.
Anthony J. Jannetti, Inc. is a provider approved by the California Board of Registered Nursing, Provider Number, CEP 5387.

Licenses in the state of California must retain this certificate for four years after the CNE activity is completed.

This article was reviewed and formatted for contact hour credit by Rosemarie Marmon, MSN, RN-BC, NE-BC, Anthony J. Jannetti, Inc., Education Director; and Judy A. Rollins, PhD, RN, Pediatric Nursing Editor.
Tyrosinemia Type 1: An Overview of Nursing Care


Pediatric Nursing Guidelines for Authors

Writing for Pediatric Nursing: This journal’s purpose is to reflect trends, policies, practices, and research in pediatric nursing. Topics should be timely, controversial, and currently unavailable in the literature. A query letter is requested, including an abstract of the manuscript and the anticipated submission date. Un-solicited manuscripts are welcome, provided that they are for the exclusive use of Pediatric Nursing and have not been previously published or accepted for publication, or are under consideration elsewhere. Authors are encouraged to use clear, concise, nondiscriminatory language.

Manuscript Form: Manuscripts should be typewritten, double-spaced, on one side of 8-1/2 x 11 inch white paper; maximum length 20 pages (5,000 words). A cover page should include the manuscript title (10-12 words); authors’ names, credentials, and primary affiliations; and the address, telephone numbers, and fax number of the primary author. This page should be followed by a substantive abstract of no more than 250 words. Manuscript title should be repeated on the first page of the text.

Figures/Tables: Tables and figures must include detailed legends. If the table or figure is taken from another source, include a full reference citation at the bottom of the page. Written permission for both print and electronic use must be obtained from the original publisher prior to the manuscript’s submission. Photographs can be color or black and white, with a high resolution (at least 300 dpi).

References: References, photographs, figures, tables, and all other details of style must conform with the Publication Manual of the American Psychological Association (APA, 6th ed., 2010). All references in text should be cited by author and date, for example, (Doe & Brown, 2010). List all references in alphabetical order.

Only use references that are actually cited within the text. Authors are encouraged to provide the digital object identifier (DOI) number directly after all references when possible. Citing multiple authors: In-text citations with six or more authors should include the first author followed by et al., even in the first citation. Seven authors or less: In the Reference section, list all seven authors: Doe, J.R., Brown, M.S., Trent, J.R., Michaels, M.S., Bradley, J.R., Willis, M.S., & Williams, J.R. (2010). Pediatric nursing care. New York: Academic Press. (Book)

Eight or more authors: In the Reference section, list the first six authors, then an ellipsis, then the last author: Doe, J.R., Brown, M.S., Smith, J.R., Jones, M.S., Thomas, J.R., White, M.S., ... James, J.R. (2010). Coping with hospitalization. Pediatric Nursing, 21(2), 115-120. (Journal Article)

Manuscripts must NOT contain reference software codes, and the use of reference software is highly discouraged.

Review Process: Receipt of manuscript is acknowledged by the editorial coordinator. Pediatric Nursing is a refereed journal; therefore, each manuscript is reviewed by members of the Manuscript Review Panel as well as the editors. Because manuscripts are reviewed blind (authors anonymous), names of authors should appear only on the cover page. Decisions regarding acceptance for publication are based on the recommendations of the referees.

Editing: Pediatric Nursing reserves the right to edit all manuscripts according to its style and space requirements and to clarify content. Edited copy will be returned to the primary author for approval.

Publication: Authors will be notified of a manuscript’s acceptance usually within 12 weeks of receipt, with publication in the next available issue. Authors may purchase reprints of their articles at the time of publication. All authors of published articles will receive 10 complimentary copies (to be distributed to any co-authors) in appreciation of their work.

Submission Procedure: Submit via e-mail one electronic copy (MS Word format only) to Editorial Coordinator Joe Tonelli at joe@ajj.com. Do not submit your manuscript as separate files (i.e., Title Page, Author Information, Abstract, etc.); combine all portions where able. Submission of hard copy is no longer required, but Pediatric Nursing defers to author preference.

Copyright: Copyright on all published articles will be held by Pediatric Nursing. Each author of a submitted manuscript signs a statement expressly transferring copyright in the event the paper is published. This form will be sent by the editorial coordinator when receipt of the manuscript is acknowledged. Authorization to photocopy items for internal or personal use, classroom use, or external or personal use of specific clients, is granted by Jannetti Publications, Inc., provided the appropriate fee is paid directly to Copyright Clearance Center, 222 Rosewood Drive, Danvers, MA 01923 USA; (978) 750-8400. For those organizations that have been granted a photocopy license by CCC, a separate system of payment has been arranged. Special arrangements for permission are required from the publisher for any other purpose.

Judy Rollins, PhD, RN
Pediatric Nursing
East Holly Avenue/Box 56
Pitman, NJ 08071-0056
8/13
Tyrosinemia Type 1
continued from page 68


Additional Readings


