INTRODUCTION

• Clinical Research Nurses (CRNs) at NIH practice under a model of care encompassing 5 dimensions
• This model of care provides a framework for the CRN to support the unique care requirements of research participants through care coordination, human subject protection, and specialized clinical expertise
• The role of the CRN requires a skill set and knowledge base essential to the care of patients with rare disorders
• CRNs operationalized the model of care for an child admitted with acute symptoms of Nephropathic Infantile Cystinosis (NIC) in the spring of 2011

BACKGROUND

• NIC is a rare, autosomal recessive disorder characterized by faulty transport of the amino acid, cystine across lysosomal cell walls
• Most common cause of inherited Fanconi syndrome in children, which causes the majority of clinical symptoms
• Cystine crystals accumulate and damage tissue in the kidneys, eyes, brain, bones, muscles, and pancreas
• Early diagnosis and treatment with the cystine depleting drug, cysteamine (Cystagon®), are essential to prevent tissue damage and optimize outcomes
• Disease complications can include blindness, fractures, diabetes, hypothyroidism, life-threatening electrolyte disturbances, and kidney failure

CASE

• A 2 year-old Bolivian child was diagnosed with NIC 5 months prior to her arrival at NIH
• Parents consented child to cystinosis natural history protocol
• Patient required urgent admission to 1NW pediatric unit for stabilization and initiation of Cystagon® treatment

PROBLEMS

• Clinically unstable patient due to acute metabolic acidosis, hypotonic renal dehydration, and hyperkalemia
• Frequent emesis, often resulting in expulsion of naso-gastric tube
• Acquired behavioral patterns: excessive water demand with refusal of other oral nutrition, resulting in inconsolable crying and vomiting if denied
• Malnutrition (<3rd percentile for weight/length for age), stunted growth, & borderline wasting
• Hypotonia and marked developmental delays- not walking or talking
• Rickets and bilateral hip dislocation

INTERVENTIONS

• CRNs assisted with the consent process & ongoing education of family related to the clinical research protocol
• Frequent laboratory monitoring and adjustment of intravenous electrolyte therapy
• In collaboration with physician and nutritionist, naso-gastric tube feeds were slowly advanced while administering intravenous sodium chloride infusion for resolution of hypernatremia associated dehydration
• Developed customized form (data collection document) and taught the parents to document water and nutrient intake as well as weighing of diapers for output estimations
• CRNs initiated a multi-disciplinary care conference which included: the research team, orthopedic surgery, endocrinology, rehabilitation medicine, nutritionist, social work, pediatric medical and nursing teams
• Taught parents proper care of newly placed percutaneous gastric tube

MODEL OF CARE

• Coordinated and collected research specimens
• Provided nursing leadership within the interdisciplinary team
• Coordinated interdisciplinary meetings & activities related to clinical care and the research protocol

OUTCOMES

• Patient stabilized from acute metabolic state
• Cystagon® treatment initiated and tolerated
• Excessive water intake decreased and nutrient intake maximized
• Growth: BMI improved to 95 percentile at 6 month follow-up
• Naso-gastric tube switched to gastric tube
• Developmental advances: walking, running, playing, talking
• Attending feeding clinic
• Family knowledgeable about condition & clinical research protocol

ACKNOWLEDGEMENTS

• Patient and Family
• Galina Nesterova, MD, Investigator for Cystinosis study
• 1 North West Nursing Staff
• Multi-disciplinary team
• Michael Krumlauf, BSN, RN, OCN
• Permissions obtained from family for photos and poster content