5-2019

Caring for the Pediatric Neurofibromatosis Type-1 Patient: Improving Nursing Knowledge through an Innovative Educational Tool

Samantha Ingerick
California State University, Northern California Consortium Doctor of Nursing Practice

ABSTRACT

CARING FOR THE PEDIATRIC NEUROFIBROMATOSIS TYPE-1 PATIENT: IMPROVING NURSING KNOWLEDGE THROUGH AN INNOVATIVE EDUCATIONAL TOOL

Standardized nursing education can help equip nurses with appropriate skills needed to care for certain patient populations. Pediatric neurofibromatosis type-1 (NF1) patients have unique health needs, and often seek care at institutions where no NF1 training has been provided. The purpose of this project was to explore existing nursing NF1 knowledge and knowledge perception in a pediatric oncology infusion center within a large bay area children's hospital, a location where nurses have varied NF1 education and have recently been asked to care for this population. Malcolm Knowles' adult learning theory was used to design an NF1 educational tool for nurses. The educational tool serves as a standardized reference from which nurses can familiarize themselves with the genetic nature of NF1, the clinical needs of the disorder, and the NF1 nursing role. A pre- and post-test survey was used to measure nursing knowledge and nursing knowledge perception, which was administered before and after the educational tool. Findings from this research suggest that the NF1 education tool is effective in improving nursing knowledge and knowledge perceptions about the complex care needed for NF1 patients. While fulfilling the American Nurses Association genetic and genomic nursing competency requirements, this educational tool can be used to standardize NF1 nursing education in hopes to enhance nursing practice and ultimately improve NF1 patient outcomes.

Samantha Ingerick
May 2019

CARING FOR THE PEDIATRIC NEUROFIBROMATOSIS TYPE-1 PATIENT: IMPROVING NURSING KNOWLEDGE THROUGH AN INNOVATIVE EDUCATIONAL TOOL

by

Samantha Ingerick

A project

submitted in partial

fulfillment of the requirements for the degree of

Doctor of Nursing Practice

California State University, Northern Consortium

Doctor of Nursing Practice

May 2019

APPROVED

For the California State University, Northern Consortium
Doctor of Nursing Practice:

We, the undersigned, certify that the project of the following student meets the required standards of scholarship, format, and style of the university and the student's graduate degree program for the awarding of the Doctor of Nursing Practice degree.

Samantha Ingerick
Project Author

Ruth Rosenblum, DNP, RN, PNP-BC, CNS (Chair) Nursing

Cynthia Campen, MD Lucile Packard Children's Hospital

Heather Radtke, CGC Children's Tumor Foundation

AUTHORIZATION FOR REPRODUCTION

OF DOCTORAL PROJECT

X I grant permission for the reproduction of this project in part or in its entirety without further authorization from me, on the condition that the person or agency requesting reproduction absorbs the cost and provides proper acknowledgment of authorship.

_____ Permission to reproduce this project in part or in its entirety must be obtained from me.

Signature of project author: *Samantha Ingerick*

ACKNOWLEDGMENTS

I wish to thank the many teachers who were involved in my academic achievements. My academic team involved in this project, Dr. Ruth Rosenblum, Dr. Lisa Walker-Vischer, Dr. Cynthia Campen, Dr. Paul Fisher, and Heather Radtke, CGC provided me with exceptional guidance and encouragement. Their time and patience helped me advance my educational and professional goals, and for these, I will always be grateful.

And to my husband, Chad: together we make each other better. Thank you for being by my side as I accomplished my goal. Let's continue to achieve great things and see how far we can go.

TABLE OF CONTENTS

Page

LIST OF TABLES

Page

LIST OF FIGURES

Page

CHAPTER 1: INTRODUCTION

According to the National Institute of Health (NIH), neurofibromatosis type-1 (NF1) is an autosomal dominant disorder that affects 1 in 3,000 people worldwide (NIH, 2016). It is the most common of the three types of neurofibromatosis, with type-2 (NF2) affecting 1 in 40,000 people and schwannomatosis affecting less than 1 in 40,000 people (NIH, 2016). NF1 can be inherited from a parent or can result from a spontaneous gene mutation (NIH, 2016), and is caused by a mutation in the *NF1* gene that is responsible for making the protein called neurofibromin (NIH, 2018). The NF1 gene is located along the long (q) arm of chromosome 17 at position 11.2 (NIH, 2018), and acts as a negative regulator along the Ras signal transduction pathway (National Center for Biotechnology Information, 2018). Abnormal neurofibromin production can lead to uncontrolled cellular activity along nerves, and ultimately form tumors under the skin surface, near the spinal cord, or along nerves located elsewhere in the body (NIH, 2018).

NF1 is most commonly characterized by other associated conditions as well. These include skin discolorations (café-au-lait spots and freckling of the axilla or groin), neurofibromas (benign growths underneath the skin), softening of bones, cognitive conditions and learning disabilities, Lisch nodules of the eyes, optic gliomas, and hypertension (Children's Tumor Foundation, 2016). The number of conditions a patient presents, and the severity of each condition, can vary between patients. Because the *NF1* gene is so large (60 exons), and because it has one of the highest rates of spontaneous mutations in the human genome, a large array of mutations can lead to the NF1 phenotype and thus the variety in phenotypic presentation (Boyd, Korf, & Theo, 2009). The Children's Tumor

Foundation (CTF) states that sixty percent of NF1 cases present with mild NF1 manifestations (CTF, 2016), while 15% of NF1 patients have symptoms that are debilitating (NIH, 2016). The NIH Consensus Development Program states that two or more of the following must present in order for a clinical NF1 diagnosis to be made: six or more café-au-lait macules over 5mm in size (pre-pubertal) or over 15mm (post-pubertal), two or more neurofibromas or one plexiform neurofibroma, freckling of inguinal or axillary regions, optic glioma, two or more Lisch nodules, osseous lesion, or a first-degree relative with an NF1 diagnosis (NIH, 1987).

Because of the wide variety of NF1 clinical presentations, treatment for NF1 can vary. For patients who develop tumors or neurofibromas, surgical resections may be needed. Optic gliomas often require ophthalmological assessments, surgery, radiation, or chemotherapy, while osseous lesions, pseudoarthrosis, or scoliosis may require orthopedic interventions (NIH, 2016). In addition, varying degrees of cognitive functioning, processing speeds and attention or hyperactivity impairment can present. Fifty percent of children with NF1 have learning challenges, poor social skills and difficulty forming friendships, all supporting the need for neuro-psychological evaluations and school needs assessments (CTF, 2016). The complex medical needs of NF1 patients often require that several medical specialists be involved. It is common for NF1 patients to be cared for by neurologists, dermatologists, cardiologists, geneticists, ophthalmologists, orthopedists, psychologists, oncologists, and school educators (CTF, 2016). Nurses play key roles in the interdisciplinary NF1 care team including symptom management, prevention of complications, ensuring family centered care and education (Sampson, Thompson, & Wall Parilo, 2019). While many nurses that care for pediatric NF1 patients and other genetic disorders are familiar with managing these complex needs, they sometimes lack experience or

have no formal educational training. As Camak (2016) demonstrated, practicing nurses lack genomic literacy, knowledge, and skills resulting in inadequate ability to meet the needs of patients and families facing genetic disorders. In 2018, Calzone, Jenkins, Culp, and Badzek found that of the 3,880,000 nurses in the U.S., most have had no genomic education (Calzone et al., 2018).

Background

With the publication of the Human Genome Project in April 2004, accurate gene sequences for each human chromosome were generated (National Human Genome Research Institute, 2016). The project continues to launch subsequent discoveries about second-generation sequencing technologies that help advance the genomics field (Hood & Rowen, 2013). Molecular medicine and genome research have since produced new information about genes involved in inherited disorders, genetic diseases and overall human health (Lessick & Anderson, 2000). Now, after nearly two decades of genetic health advancement, there is the challenge of maintaining a competent workforce that can adequately translate these genomic discoveries into practice (Calzone et al., 2018). Genomic information continues to transition into the clinical setting at quick rates, creating an urgency for medical professionals to receive genetic disorder education and provide competent care (Calzone et al., 2018). Nurses are included in this group of health professionals who have a responsibility to be knowledgeable about genetic practices and to incorporate scientific advancement.

The Genetic/Genomic Nursing Competency Initiative (GGNCI) was formed in 2004 from collaboration between the National Human Genome Research Institute and the National Cancer Institute (Genomic Nursing State of the Science Advisory Panel, 2013). This group prioritized efforts to create a

genomics/genetics nursing competency strategic implementation plan. The plan aimed to recognize that genetic nursing knowledge can impact patient care during risk assessments and discussions surrounding treatment decisions (Genomic Nursing State of the Science Advisory Panel, 2013). This partnership, in collaboration with the American Nurses Association (ANA) produced a document titled *The Essentials of Genetic and Genomic Nursing: Competencies, Curricula Guidelines, and Outcome Indicators* published in 2006, and later revised in 2008. This document aimed to reflect the minimal amount of genomic and genetic competencies expected from every nurse (ANA, 2009). It was funded by the NIH, the National Human Genome Research Institute, the Office of Rare Disease, and the ANA (Lewis, Calzone, & Jenkins, 2006). This publication was agreed upon by a consensus of 47 endorsing organizations including two schools of nursing and members of the Nursing Organization Alliance (Lewis et al., 2006).

The document provides definitions of basic genetic and genomic terminology, professional nursing responsibilities with regards to genetic health, and expected outcomes for each competency. It includes implementation strategies that outline the basic nursing educational requirements needed so that graduating nurses could deliver adequate genomic and genetic care (ANA, 2016). These guidelines were intended to apply to all registered nurses regardless of academic preparation, practice setting, role or specialty and aim to prepare the nursing workforce to deliver competent, genetic and genomic focused nursing care (Lewis et al., 2006). This document has since been considered the gold standard for practicing nurses regarding genetic and genomic care (ANA, 2016).

Also in 2008, the American Association of Colleges of Nursing (AACN) published an updated version of the *Essentials of Baccalaureate Education for Professional Nursing Practice* that outlined curricular elements and frameworks

used as guidelines for nursing educational programs around the U.S. (AACN, 2008). Understanding that nursing has potential to greatly impact healthcare delivery to patients, the AACN created 9 curriculum outcomes expected of baccalaureate nursing graduates that emphasize patient-centered care, evidence-based practice, quality improvement, genetics and genomics, cultural sensitivity, professionalism, and other skills (AACN, 2008; Connors and Schorn, 2018). In 2011 the AACN acknowledged a subsequent report from the ANA and International Society of Nurses in Genetics (ISONG) titled *Essential Genetic and Genomic Competencies for Nurses with Graduate Degrees* (AACN, 2011). This builds on the original 2009 consensus panel document while incorporating genetic advances and genetic competency guidelines for nurses with higher level degrees and leadership roles. In addition to these institutions, other organizations have put forth efforts to standardize genomic and genetic nursing education (see Appendix E).

Despite these many organizations attempting to regulate the amount of genetic-related education received by nursing graduates, a gap in genomic knowledge continues to exist amongst registered nurses today. This could be the result of many reasons. Calzone et al., (2018) explain that the complexity of genomic concepts, the ability of nursing academic institutions to change curricula, and the lack of state boards requiring genetic competencies as part of licensure may all play a part in the sluggish integration of this skill into the nursing culture. The National Council of State Boards of Nursing (NCSBN) Test Plan for 2019 claims to devote 6-12% of NCLEX Test Plan questions to the health promotion and maintenance category, which is divided into 9 subcategories. One of these subcategories contains questions related to genetic screening, history, or risk assessment (NCSBN, 2018). While it is difficult to determine the exact number of

test questions pertaining to genetic or genomic content, it is likely to be a small percentage of the exam.

Transforming these genetic advances into current nursing practice can also be challenging for nurses (Camak, 2016). With recent NF1 genetic discoveries emerging, nurses are asked to maintain a sufficient grasp of core scientific NF1 concepts (Camak, 2016). Considering the prevalence and complexity of NF1, it is likely that nurses will encounter patients with NF1 at some point throughout their careers (Julian, Edwards, DeCrane, & Hingtgen, 2014). Those nurses with a generalized sense of current NF1 education may be better equipped to identify children with NF1, obtain a family history, conduct pertinent physical and development assessments, and provide the family with basic genetic information and psychosocial support (Lessick & Anderson, 2000). A nurse who understands the pros and cons of NF1 genetic testing may be able to serve as an advocate for the patient, discuss screening or treatment choices, and contextualize any potential risks (Camak, 2016). Genomic knowledge gaps can lower the effectiveness of utilizing genomic information during health care decision-making. This can affect patient safety and outcomes of care (Calzone et al., 2018). In response to these gaps, efforts have been made to educate professionals about NF1 and the clinical skill set needed to provide optimal care.

The CTF is a national nonprofit foundation that was founded in 1978 to help find treatments for NF1 (CTF, 2016). The CTF created the Neurofibromatosis Clinic Network (NFCN) in 2007 to help standardize NF1 care and integrate research into clinical practice within the U.S. Currently 50 NF clinics within the U.S. are registered as specialty care clinics for this population and are dedicated to providing comprehensive medical care to those with NF, fostering patient education, promoting support and enrollment for NF1 clinical

trials, and updating the national NF patient registry (CTF, 2016). The Neurofibromatosis Network is another non-profit 501(c)(3) organization based in Illinois that hopes to share resources, research, and improvements in clinical care in attempt to find a cure for neurofibromatosis and optimize patient outcomes (Neurofibromatosis Network, 2018). Neurofibromatosis Inc. California is also a non-profit, volunteer organization located throughout California that was founded in 2004. It is composed of individuals and families affected by NF1, and provides educational forums, family support groups, and resources for healthcare professionals and patients (Neurofibromatosis California, n.d.). These organizations share common goals dedicated to advancing NF1 research, to educating others about NF1, and strive to supporting patients and families living with this disorder.

Role of the NF1 Nurse

When providing medical supervision to a child with NF1, the nurse or care coordinator plays a unique role. Because of the various NF1 phenotypes, different NF1 patients may need to be monitored in various medical departments. For example, some NF1 patients may be followed by genetics physicians and nurses, while other NF1 patients may be seen in the neurology or dermatology department. The nurses in each of these departments all share the responsibility of providing competent care outlined by the ANA and the AACN to emphasize health promotion, prevention, caring, screening, and relationships (Munroe & Loerzel, 2016).

Since the *Scope and Standards for Clinical Genetics Nursing Practice* was published in 1998 by the ANA and the International Society of Nurses in Genetics, genetics has been recognized as a nursing specialty (Montgomery, et al., 2017).

Learning outcomes were revised after the Human Genome Project was completed, and the ANA published the *Essentials of Genetic and Genomic Nursing: Competencies, Curricular Guidelines, and Outcome Indicators* in 2009. From these documents emerged preliminary descriptions of the nursing role when caring for patients with genetic disorders, and they specifically outlined genetic-related nursing skills expected of all nursing school graduates in the U.S. (Montgomery et al., 2017). The nursing competencies listed in the ANA documents are divided into two categories: professional responsibilities and professional practice. Professional responsibilities include recognizing one's attitudes related to genomic science that may affect client care, or advocating for the rights of all clients for autonomous, and informed genetic-related decision-making and voluntary action. Similarly, professional practice includes the ability to elicit a 3-generation family history, developing a plan of care that incorporates genetic assessment information, identifying patients who may benefit from specific genomic information or services, and providing patients with knowledge of genetic-related risk factors or disease prevention practices (ANA, 2008).

Since the development of these guidelines, the expectations of nurses caring for patients with genetic conditions have not changed. The nursing workforce is expected to utilize a genetic pedigree while developing care plans, deliver patient education, and even provide some genetic counseling, referrals, or psychosocial health assessments (Camak, 2016). With regards to the NF1 population, nurses have the task of familiarizing themselves with NF1 so as to provide the patient and family comfort. Barke, Coad, & Harcourt (2016) conducted a qualitative study in England that explored parents' experiences of caring for a child with NF1. Parents described feeling frustrated and angry because health professionals had not heard of NF1, or misunderstood it. Results

from this study highlighted the value that parents place on up-to-date NF1 information, access to health care professionals with NF1 knowledge, and on the importance of general public NF1 awareness. It is important that the NF1 nurse addresses psychosocial needs of patients and families to provide them with comfort and reassurance. The nurse or care coordinator role includes providing proficient genetic and genomic care, but also involves translating NF1 knowledge into competent and confident nursing care so that families feel supported.

Significance

The birth incidence of NF1 is one in 1,900 to 2,800 cases worldwide. The diagnostic prevalence, however, is higher (one in 4,150 to 4,950) because of the cases that are discovered later in early childhood, adulthood, or even at time of death (Evans, et al., 2017). When looking at 20 year olds with NF1, a retrospective review study found that only 54% of them met criteria at age 1, 97% of them met criteria at age 8, and 100% met criteria by age 20 (Boyd, Korf, & Theos, 2009). About 50% of NF1 cases result from spontaneous mutations and 50% are inherited from a parent (Rasmussen & Friedman, 2000). California is no exception to this prevalence, and with 1,938,153 people in Santa Clara County alone, any level-1 pediatric trauma children's hospital in that area would be expected to encounter patients who require NF1 care (US Census Bureau, 2017).

One particular children's hospital in the California bay area has recently joined the NFCN and become active within the CTF group to enhance the pediatric NF1 care within the Bay Area of Northern California. Nurses within the pediatric oncology infusion center (a department within the larger hospital) are now being asked to care for NF1 patients. While the pediatric oncology nurses most likely had prior exposure to the pediatric NF1 population, their NF1

knowledge, education, and experiences are unknown and varied. In preparation for transitioning the pediatric NF1 patients to the infusion center, an opportunity presented to examine the existing NF1 knowledge and nursing NF1 knowledge perceptions of these nurses. Further investigation looked into the existence of a standardized nursing NF1 educational tool that could educate infusion center nurses should they be unfamiliar. A novel NF1 educational PowerPoint tool was thus created and examined to determine its effectiveness in improving nursing NF1 knowledge and knowledge perception.

Problem Statement

There currently exists no standardized educational tool shown to improve nursing knowledge and nursing knowledge perceptions of pediatric NF1 care despite regulatory genetic and genomic educational requirements of the AACN and the ANA (AACN, 2008; ANA, 2008).

Purpose of the project

The purpose of this project was to create an effective educational tool (PowerPoint) for nurses to help improve nursing knowledge and nursing knowledge perceptions of pediatric NF1 care. By measuring nursing knowledge of pediatric NF1 care before and after using this educational tool, the tool's effectiveness could be determined. In attempt to follow the AACN nursing educational guidelines and equip nurses with the academic background to provide safe and appropriate care for all patients with genomic and genetic conditions (AACN, 2008), the NF1 educational tool was made to help meet these goals. The intentions of this tool also incorporated the mission of the CTF, which is to drive research, expand knowledge, and advance care for the NF community (CTF,

2016). In doing so, this project aimed to benefit nurses' NF1 education and ultimately improve NF1 patient outcomes.

CHAPTER 2: LITERATURE REVIEW

Existing Evidence

To date, a standardized nursing NF1 educational tool that teaches the essential genetics and genomics curriculum requirements stated in the AACN does not exist (AACN, 2008). While literature devotes attention to educational tools, websites, curricula guidelines and simulations pertaining to unspecified genetic disorders (ANA, 2008), none of these are specific to NF1 patients. Knowing that NF1 patients require unique care that incorporates medical, psychosocial, and academic needs, there is an apparent void of standardized learning tools intended to help medical professionals learn about NF1. Comprehensive literature searches using the full-text database Nursing and Allied Health search engine provided by the Henry Madden Library at University of California, Fresno State offered several peer-reviewed articles that highlight the importance of nursing genomic and genetic knowledge and competence. While the literature suggests a disconnect between the expectations of nursing genetic competencies and the actual skill level portrayed in the nursing workforce, there seems to be no suggestion of a nurse-specific tool that could help resolve this disconnect. The following literature review examines current publications that highlight the existing evidence surrounding nursing genetic competency and proficiency, as well as the lack of literature pertaining specifically to pediatric NF1 nursing knowledge.

Camak (2016) conducted an extensive literature review looking at 20 journal submissions from 2008 to 2015 relating to the incorporation of genetics into nursing practice. Databases used included CINAHL, PubMED, American Association of Colleges of Nursing, NCHPEG, International Society of Genetic

Nurses and ProQuest Nursing. Of the 20 articles retrieved, 6 were research articles, 4 were published documents, and 10 were informational articles. Camak's results found a consistent trend indicating that nurses lack genetic competency. The author quotes Anderson et al. (2015), "despite a large and ever growing field of genomics knowledge, the profession of nursing has not yet incorporated core competencies for genomics into annual RN competency assessment and evaluation." Results suggest that barriers to integrating genetic content into nursing practice include poor understanding of its relevance, lack of state boards requirements for nurses to grasp this training prior to licensure, and limitations that prevent nurses from interpreting scientific genetic information and applying it to patient care.

To help determine if nurses were utilizing elements of the AACN's *Essential Nursing Competencies and Curricula Guidelines in Genetics and Genomics (Essentials)* in daily practice, Thompson and Brooks (2011) conducted a cross-sectional survey study. The 17-question survey was sent to 200 nurses recruited from a convenience sample of conference attendees. The survey included questions regarding nursing curriculum content, continuing education, nursing certification, and involvement in genetic/genomics research. The survey had a 24% response rate (n=47), and of those respondents only 36% had read the *Essentials* document. Thompson and Brooks' (2011) results suggest that content from the *Essentials* had not been reviewed by nurses other than those actively involved in genetic research or genetic continuing education, and that most respondents claimed that their school of nursing did not fully meet the Essentials competencies. While this study had a small sample size and skewed results considering its subjects were recruited from a conference and may be academically focused, it concludes that most nurses did not have sufficient

knowledge regarding genetics and were not familiar with the competencies in the AACN *Essentials* document.

Calzone, et al.(2018) were interested in exploring the effectiveness of a year-long program aimed at improving nursing ability to translate genomic information into clinical practice. This longitudinal study took place from 2012 and 2013 at 23 hospitals: a control group (2 hospitals) and an intervention group (21 hospitals representing 14 states). Routine nursing education was offered monthly at the intervention hospitals only. The Genetics and Genomics Nursing Practice Survey (GGNPS) was administered to nurses pre-intervention and post-intervention (n=8,150 RNs). Results indicate that intermittent nursing educational interventions such as genomic awareness campaigns, and personal genomic competency endorsement can increase nurses' adherence to guidelines when integrating genomics into nursing practice. It also found the long intervention (1 year) design improved genomic competency in the participating nurses, and that having a leadership involvement increased the likelihood that nurses would engage in learning to apply genomic information to bedside practice.

Munroe and Loerzel (2016) created a pre-test/post-test survey study using a convenience sample of 120 baccalaureate nursing students at a nursing school in Florida (n=120). The surveys were administered before and after a semester in which they would receive heavy genetics academic content. The Genomic Nursing Concept Inventory (GNCI) involved 31 multiple choice questions covering topics such as Human Genome Project, mutations, inheritance patterns, genomic health care applications, and attitudes about using genetic information in practice. Results indicated a significant relationship (r=0.22, P=.02) between knowledgeable students and positive attitudes about their ability to use that knowledge. Knowledge gains were seen between the pre-test and post-test, with a

difference of only 5% however. Most students felt responsible for knowing the material covered in the course, but did not feel ready to practice this material with clinical patients. This study suggests that students need more exposure to genetic conditions, possibly through re-evaluation of nursing curricula that can leave students feeling more confident in genomic care.

Draucker, Nutakki, Varni, and Swigonski (2016) conducted a qualitative semi-structured interview-style study that utilized the Pediatric Quality of Life Inventory (PedsQL) NF1 model. This tool helped collect comprehensive descriptions from pediatric NF1 patients and families, pediatric participants were divided into age groups (5-7yrs, 8-12yrs, 13-17yrs, and 18-25yrs) and interviewed in person while parents of these participants were interviewed via phone (n=41). Semistructured and open-ended questions were used to elicit narratives regarding how NF1 affects physical symptoms, treatments, psychological and school functions. Narratives were tape-recorded and transcribed. Results suggest that several NF1 patients and families have to explain NF1 to others because it is poorly understood in the community. Several participants mentioned having a number of worries about NF1, mostly about the uncertainty of it progressing. The authors created a framework to include the five most important concerns from participants: pain, social functioning, physical limitations, stigma, and emotional distress. This study highlights the need for frequent quality of life nursing assessments for NF1 patients so that these concerns can be addressed and supported.

Baker (2011) conducted a cross-sectional survey design study in the UK that examined 368 nurses (n=368) and used a 30-item, anonymous questionnaire. The survey was distributed to all nurses hired at a specialist hospital during 2008,

and was sent to the nurses' homes in the public mail. Subjects had 5 weeks to complete the questions and return the survey in a previously stamped envelope. 26 questions were closed statements, and 4 questions were open-ended. 91% of respondents claimed that nurses require access to specialist education and training that focuses on neurological conditions. Nurses often perceived that they were ill prepared for practice in any setting or specialty. This study raises the possibility that nurses in the U.S. may be experiencing similar degree of preparedness towards NF1 patients, and would gain confidence if additional training were provided.

Chen and Kim (2014) developed a survey to assess the genomic education training needs among health educators. The subject sample was formed by purchasing a list of people who were Certified Health Education Specialists (CHES), which is a certification granted by the National Commission for Health Education Credentialing, Inc (NCHEC). 7,626 health educators with CHES designation were invited to take part in this study (n=7,626) and 980 health educators chose to participate. The questionnaire included questions items regarding previous training in genomics, self-reported genomic knowledge, beliefs and values of incorporating genomics into health promotion and practice, desired genomic training, and preferred delivery methods. 60.6% of respondents claimed they had no or very little genomic knowledge. 5.6% stated to have quite a lot or an extensive amount of knowledge. The preferred methods for genomic training and education were (in order of popularity) continuing education, web-based training, professional conferences, workshops, interpersonal communication, peer-reviewed articles, in-service training, and teleconferences. This study had low response rate (12.9%) and those that participated might be biased towards learning about genomics training compared to others who did not participate. These might

have limited the study's findings, but was able to conclude that overall there is a need for genomic education even amongst health educators in the U.S.

Gallo, Angst, Knafl, Twomey, and Hadley (2010) set out to examine the views of health care professionals regarding how to care for patients and families with genetic disorders. Semi-structured interviews were used to collect information from 37 health professions in 3 clinical sites in the midwest, U.S. A goal of this study was to see how health professionals are individualizing care and genetic information for patients with genetic disorders. Professionals were recruited for this study after they were recruited to partake in a larger study involving parents. Eight registered nurses took part in these interviews (22% of participants), and they all had some experience with either phenylketonuria, sickle cell disease, cystic fibrosis, neurofibromatosis, hemophilia, thalassemia, Marfan syndrome, or von Willebrand disease. Results conveyed four major themes: (1) sharing information with parents, (2) taking into account parental preferences, (3) understanding of the condition, and (4) helping parents inform others. The nurses emphasized their role of reinforcing information from the physicians, but also concentrating on care coordination, clinic resources, and anticipatory guidance education. One nurse mentioned that parents are overwhelmed at times, and it "is our role to help them process things they get from outside." Another nurse states that he or she is involved in translating genetic information so that families can understand, and offering reassurance when interpreting a diagnosis. While this study examined the interdisciplinary efforts from physicians, genetic counselors, nutritionists, and social workers in addition to nurses, it is able to conclude that health care professionals play a central role in assisting families of children with genetic conditions in understanding the condition.

These literary examples consider several aspects of nursing preparedness, knowledge, and competency in relation to caring for patients with genetic and genomic conditions. It is clear from these articles that nurses are unfamiliar with the baccalaureate genetic essentials and competencies expected of all nursing graduates. This unfamiliarity translates to a lack of core nursing skills and a poor understanding of these patients' basic needs. In addition, the literature shows that nurses perceive themselves as being unprepared for genetic and genomic patient care and recognize their own professional deficiencies. Possibly the most compelling evidence of this shortcoming stems from the families and patients who feel that health care providers misunderstand these disorders. They state that the most frustrating part of their medical experience is when encountering providers who are unfamiliar and uneducated about their child's disease. Other literature explores ways in which genetic and genomic education can be promoted, some suggest long term interventions and having leadership involved in advocacy and endorsement. It is clear that heavy genetic academic content is shown to improve genetic knowledge, and yet there have been no proposed standardized educational tools that attempt to achieve this. The responsibility of health care professionals is to understand these conditions to the extent set forth by the AACN and the ANA, so that nurses have competencies and confidence to safely and appropriately care for these patients. The review of the literature above shows that improvements are still needed before such can be achieved.

Conceptual Framework

The nursing NF1 education project was based from the idea that adult nurses would gain knowledge about NF1 and the NF1 nursing role by using a self-guided educational tool. The theoretical/conceptual basis for this project

integrates Malcolm Knowles' adult learning theory that establishes the conditions under which adult learning can be maximized (Hartzell, 2007). In 1974, Malcolm Knowles coined the term *andragogy* to mean the art and science of adult learning (Kenner & Weinerman, 2011). He felt adults made up a distinct learning population that was unique to youth learners (a science termed *pedagogy*), and was characterized by six assumptions (Knowles, Holton III, & Swanson, 2005): (1) adults know why they need to learn something before undertaking to learn it. Thus, when educators emphasize the "need to know" to adult learners, effectiveness of teaching improves. (2) Adults have a self-concept of being responsible for their own decisions, and therefore strive to be self-directing learners rather than dependent learners. If educators can harness and engaged experience with the learners, rather than feeding them information, students will be more engaged. (3) Adult learners have lived through experiences that youth learners have not. These experiences serve as a lens through which context can be applied, and the adult learner will grasp concepts more strongly once experience has been applied. (4) Adults possess a readiness-to-learn that youth learners do not always have, suggesting that there is an importance to the timing of certain information. (5) Adult learners consider the orientation to learning, meaning that they grasp the reasoning that makes a lesson applicable to real life. And (6) adults carry an external motivation to learn new information, tasks, or skills. Typically this motivation comes from a salary promotion, better job, or improved quality of life (Knowles et al., 2005).

These assumptions from Knowles' theory of the adult learner can be applied to this subject population used in the nursing NF1 education project. Here, the participating subjects were oncology infusion center nurses with BSN degrees. The introductory consent form and the educational tool attempt to

address these six assumptions by highlighting the reasons why it is important they gain NF1 knowledge. The participating hospital plans to relocate all pediatric NF1 patients to a new department where they will receive care and care coordination. This new department, the oncology infusion center, is composed of oncology nurses with varied NF1 patient experience. As pediatric NF1 patients transition to the infusion center, nurses will understand the need for preparedness and a sense of purpose when caring for these patients. By using Knowles' theory of andragogy, this motivation will aid in the knowledge retention when reviewing the NF1 educational tool and adult learning can be maximized. The adult learning theory will have many opportunities to be used as adult healthcare professionals continue to face ever changing technology, genetic discoveries, and medical advancements, and thus continual opportunities to learn (Clapper, 2010). By understanding the conditions that maximize adult learning, not only will evidence-based practice be promoted but employee retention will improve and health care errors reduced (Clapper, 2010).

CHAPTER 3: METHODOLOGY

The nursing NF1 education project used a cross-sectional, descriptive research design that involved a pre-test survey, a self-guided educational tool, and a post-test survey. Approval to conduct this study was granted by the IRB at Stanford University and by the University of California/Fresno State. Participating subjects consented to the nursing NF1 educational project within the introductory email. Each subject was given 3 weeks to complete all three components: the pre-test, the educational tool, and the post-test. Data from the pre-test surveys were compared to data from the post-test surveys in order to determine if there was a change in nursing NF1 knowledge or NF1 knowledge perception after having completed the educational tool. After analyzing the results of this project, we anticipated the need for more substantial nursing education relating to NF1. We predicted that these findings would effectively contribute to NF1 nursing education and to the efforts of meeting the nursing competency essentials outlined by the ANA and AANC (ANA, 2009; AANC, 2008). With supporting data of its effectiveness, this tool could then be used by nurses at other institutions to improve nursing NF1 knowledge and ultimately improve NF1 patient care worldwide.

Participants

Inclusion criteria were limited to BSN prepared nurses within the oncology infusion center. Medical assistants, physician assistants, nurse practitioners, physicians and clinical nurse specialists were excluded. The infusion center nurse manager identified all infusion center nurses, which totaled 50 eligible subjects (n=50), and provided each of their email addresses. Permission was given by the nurse manager to use these email addresses, as well as permission for subjects to

participate in the project using work-hours. Within the surveys, subjects self-identified as infusion center nurses and stated their experience working with NF1 pediatric patients. The subject recruiting process used email as a way to request subject participation, and within the email was a statement explaining the subjects' participation as voluntary and that no compensation will be provided. Subjects' email addresses were not identifiable on the pre- or post-test survey responses, and therefore participation was anonymous to examiner. It was anticipated that each subject would benefit educationally from this study by having completed the nursing NF1 educational tool (PowerPoint). The intent of the study is that each subject would gain knowledge relating to the pediatric NF1 population, which would increase understanding, competency and comfort when caring for these patients. Pediatric NF1 patients seeking care at this children's hospital will benefit from this study because the NF1 nurse participants will have been exposed to NF1 education, helping to boost their NF1 care, confidence and competency. Greater knowledge of genomics and genetics will enable nurses to feel more comfortable and be more proficient in providing holistic care for patients and families with genetic conditions (Munroe & Loerzel, 2016).

Potential Risks:

Nurses asked to participate in the study may not have available time during work hours to complete the pre-test, educational tool (PowerPoint), and post-test. In this instance, nurses were encouraged to complete these components voluntarily outside of work time. Nurses that were unable to find time to participate were not used as subjects for this study. To minimize the risk of experiencing potential psychological anxiety throughout this study, subjects were informed of the voluntary and anonymous nature of the project. To ensure

confidentiality, subjects' email addresses were not associated with survey data. Once surveys had been completed, data was stored in Qualtrics electronic software without subjects' names or other identifying information, and with only the examiner having access. The IRB mandated 12 months of Qualtrics data storage, to which only the author has access. Data will be erased from Qualtrics by the examiner at the completion of this project or after 12 months, whichever occurs first. This study expires on September 22, 2019.

Sampling Procedure

Each subject was sent an individual email (see Appendix A). All emails were sent out simultaneously. The email contained a short paragraph describing the study instructions. The instructions listed four steps for the participant to follow: (1) read and sign the NF1 consent form (word document attachment, see Appendix B), (2) complete the pre-test survey (hyperlink to Qualtrics survey, see Appendix C), (3) review the Power Point educational tool (attachment, see Appendix D), (4) and complete the post-test survey (hyperlink to Qualtrics survey, see Appendix E). Subjects had the option of completing these steps from a computer or a cell phone that utilized internet and had access to work emails. The pre-test and post-test were identical, and consisted of 21 questions using Likert scale and multiple choice formats so as to collect nominal and categorical data. Subjects were able to manually click through the PowerPoint educational tool at his or her preferred pace, and data was anonymously and automatically saved in Qualtrics. The survey was accessible to the participants for 3 weeks (21 days). Reminder emails were sent once weekly during this period to help achieve higher response rates. Data collection occurred by downloading results from Qualtrics, and a consulting statistician assisted with statistical analyses. Each subject had

single-use participation which was ensured by asking identifying questions at the beginning of each pre-test and post-test (questions such as "what was the name of the street you grew up on?" and "what was the name of your first pet?"). No duplicate answers to these questions were found in any of the surveys. Subjects were not financially compensated for taking part in the research study.

Measures

The outcomes for the nursing NF1 education project were: (1) create a nursing NF1 education tool that is shown to be effective in improving nursing NF1 knowledge and nursing NF1 knowledge perceptions, (2) support the mission of the Children's Tumor Foundation to drive research, expand knowledge and advance care for the NF community (CTF, 2016), and (3) contribute to the nursing educational guidelines set forth by the AACN aiming to equip nurses with an academic background so that they may provide safe and appropriate care for all patients with genomic and genetic conditions (AACN, 2008). To meet these objectives, pre- and post-test surveys were given to 50 nurses that were to be completed before and after reviewing the NF1 educational tool, respectively. The pre- and post-test surveys were identical and contained 21 questions total. One question identified the survey as being either pre-test or post-test, one question asked for identifying information needed to associate each subjects' pre- and post-test while keeping anonymity of each subject (asked for childhood street name, and name of first pet), and three questions asked about professional experience (years working as a nurse, years working in pediatric oncology, and former NF1 education). Three questions asked subjective information relating to nurses' perceptions: rate your current NF1 knowledge (Likert scale), rate your perception on 13 various NF1 topics (Likert scale poor, good, excellent), and if you currently

feel prepared to care for NF1 patients (Likert scale). The remaining 13 questions were multiple choice questions testing for objective NF1 knowledge. See Table 1 for the breakdown in test questions. The nursing NF1 education project was conducted entirely electronically using Microsoft Outlook (email), Qualtrics (online pre-test and post-test), and Microsoft PowerPoint (NF1 educational tool).

Table 1. Breakdown of Pre-Test and Post-Test

Type of Test Question	**Number of Test Questions**	**Question Descriptions**
Distinguish pre- and post-test	1	-choose either pre-test or post-test
Anonymous subject identification	1	-street name & name of first pet
Former education (Background information)	3	-former NF education (L) -total RN experience (MC) -total pediatric oncology experience (MC)
Subjective	3	-rate current NF knowledge (L) -rate preparedness to care for NF1 patients (L) -rate perception of understanding of 13 different NF1 topics (L)
Objective NF1 knowledge	13	-diagnosis criteria (MC), 1 -types of NF (MC), 1 -genetic characteristics (MC), 2 -clinical presentation (MC), 3 -NF1 treatment (MC), 1 -NF1 care management (MC), 1 -NF1 nursing role (MC), 2 -psychosocial (MC), 2

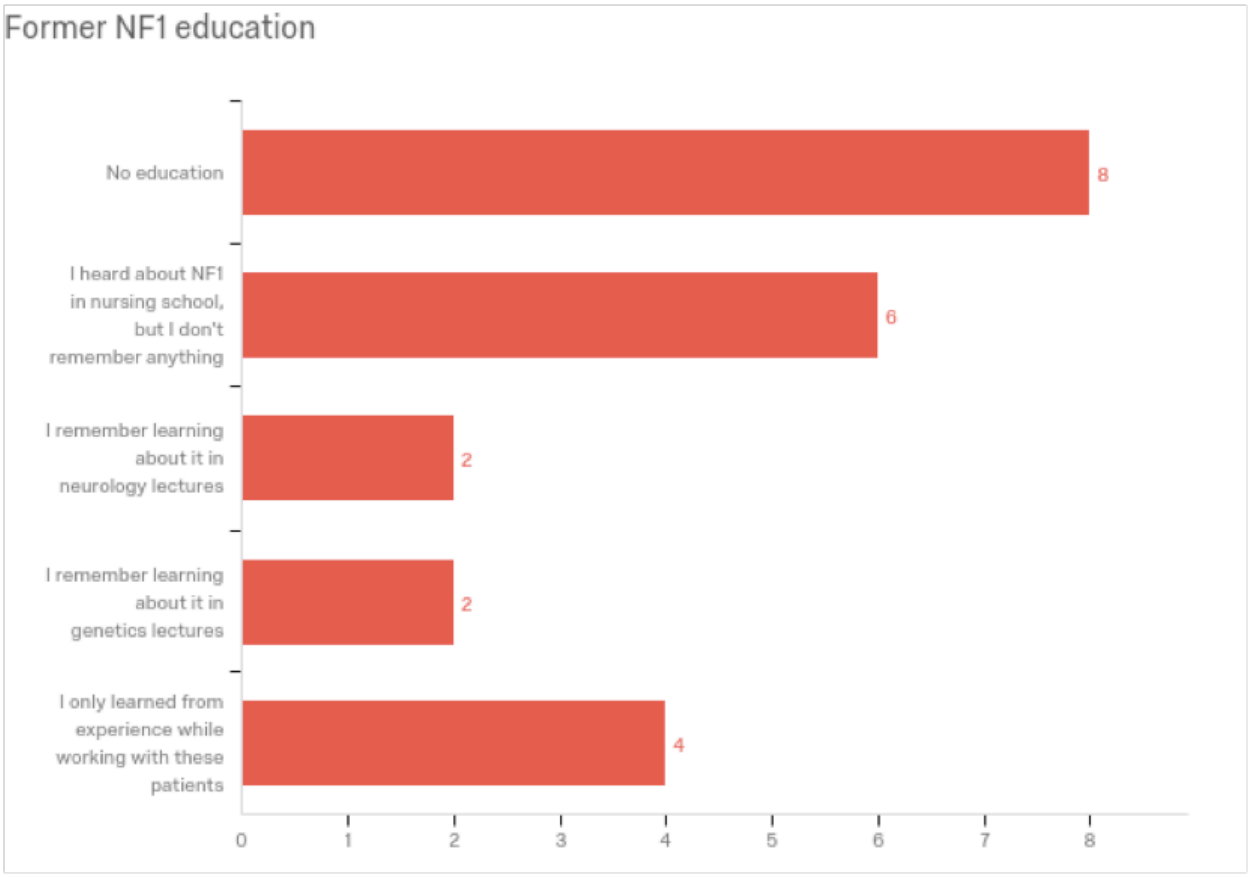
L = Likert
MC = multiple choice

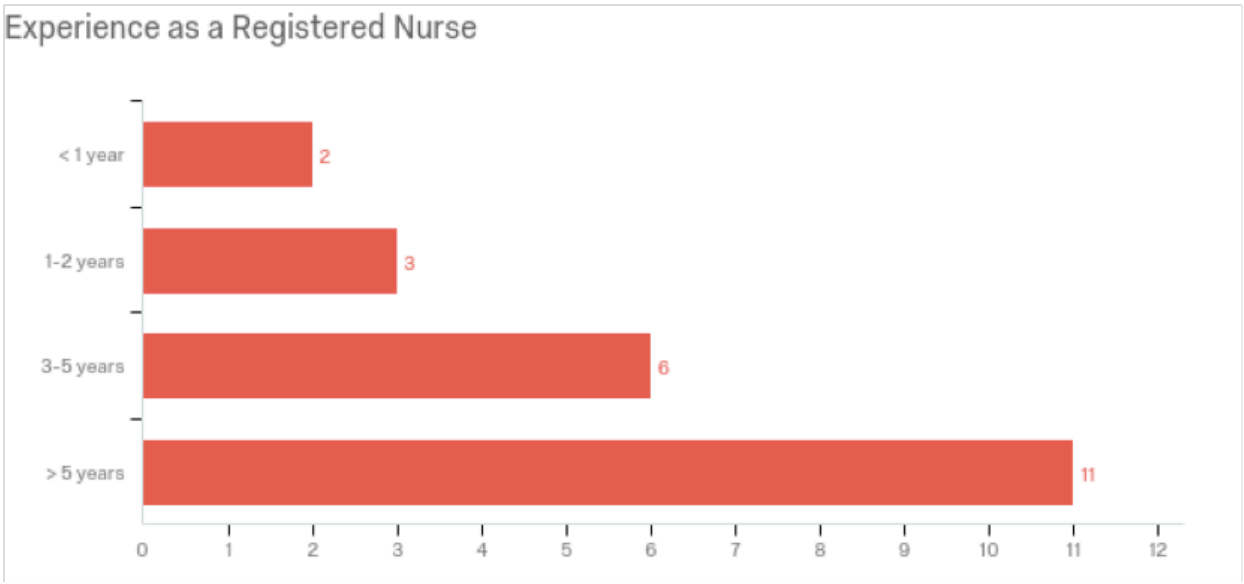
CHAPTER 4: RESULTS

Fifty infusion center nurses were asked to take part in the study by way of an invitational email. Twenty-five respondents completed the pre-test (53.19%) and 22 completed the post-test (46.81%). Only subjects that had completed both the pre-test and post-test were considered in this project, and therefore the 3 pre-tests that had no associated post-tests were discarded and not included in the statistical analysis. This resulting in a 46.81% overall response rate with n=22 (pre-test) and n=22 (post-test). This led to a small sample size.

Nursing Background Results

The majority of participants had been a registered nurses for over 5 years (50.00%), with only 2 newly graduated participants with less than one year experience (9.09%). 36% of respondents had over 5 years of experience working with pediatric oncology patients. When asked in the pre-test about the degree of former NF1 education received, 5 nurses (22.73%) stated no education, 6 (27.27%) claimed they had heard of NF1 in nursing school but don't remember anything, 4 (18.18%) remembered learning about NF1 in neurology lectures, 2 (9.09%) remember NF1 from genetics lectures, and 5 (22.73%) only learned from experience while working with these patients (see Fig. 1).





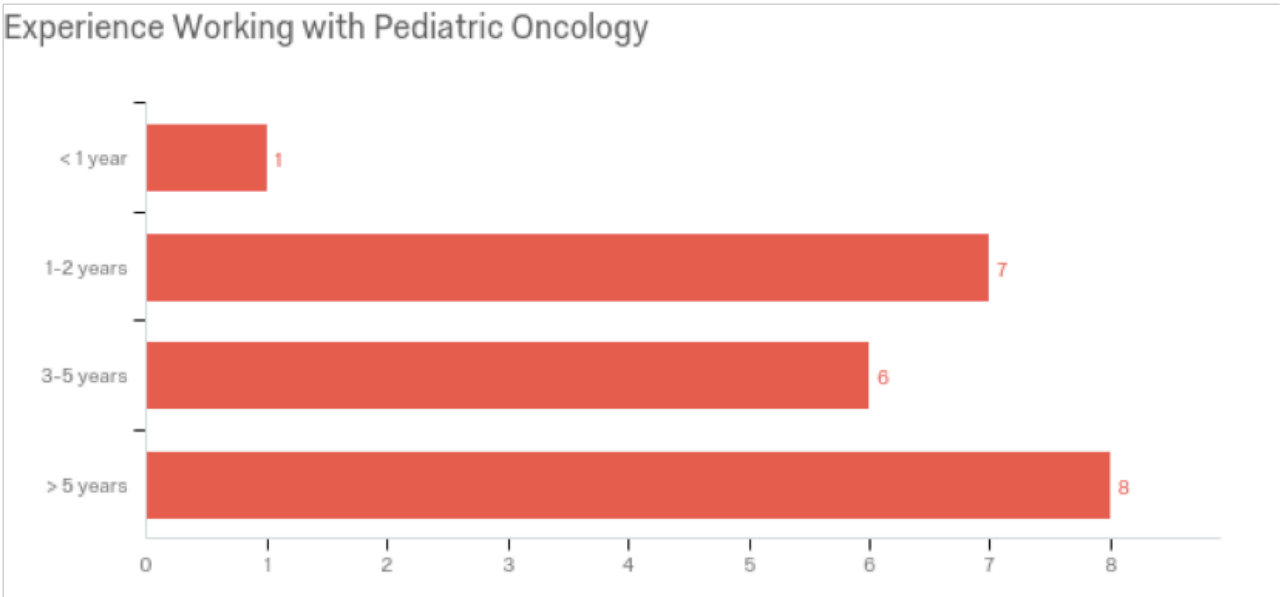


Figure 1. Nursing Background Information

Objective and Subjective Information

Twenty-two students were tested before and after NF1 educational tool training in hopes to determine if the educational tool impacted the post-test results. These tests were of two types. The first was a set of objective test questions. The second was a set of subjective test questions measuring the perception the students had in their ability and confidence in caring for pediatric patients with NF1. With a small sample size such as we have, real effects may be masked in the sense that it is difficult to statistically find effect unless they are really pronounced. However, we found that there were clear improvements (comparing the post-tests with the pre-tests) in the mean scores. For the effects of the years of experience as a registered nurse, there were no statistically significant effects in either the objective or subjective tests, whether the measurements are the initial mean scores or the improvements. On the other hand, the effects of the years of experience in pediatric oncology, while not formally statistically significant, were close to significance with regard to objective initial scores and improvement in scores.

Those with greater experience tended to have higher objective scores followed by smaller improvements. They also tended to have smaller subjective improvements. The effect of prior NF1 experience was not found to be statistically significant for either the objective or subjective tests. The correlations between the objective and subjective test scores, either the initial scores or the improvements were not found to be statistically significant.

Improvement in Scores

The objective scores are the numbers of correct answers for the 13 NF1 knowledge questions for both pre-test and post-test. We observe that the post-test had higher scores than the pre-test by an average of five points, averaged over the 22 students. We are testing that the true mean difference is zero. This is done by a paired t test. The results for the objective scores are summarized in Table 2. The t statistic is 5.54. This corresponds to a p-value of 0.000. Any p-value less than 0.05 is considered to be statistically significant. Thus, we find that the improvement is statistically significant.

Table 2. Comparison of Objective Scores, Before and After

	Before	After
Means	17.55	22.55
St. Dev.	4.09	2.94
df	21	
t	5.54	
p	0.000	

The subjective scores are the sums of all nursing perception questions, both pre-test and post-test. The results for the subjective scores are summarized in Table 3. The t statistic is 8.00. This corresponds to a p-value of 0.000. Thus, we find that the improvement is statistically significant.

Table 3. Comparison of Subjective Scores, Before and After

	Before	After
Means	15.82	26.18
St. Dev.	3.61	5.69
df	21	
t	8.00	
p	0.000	

The observed objective mean scores for both before and after are plotted in Figure 2. The observed subjective mean scores for both before and after are plotted in Figure 3.

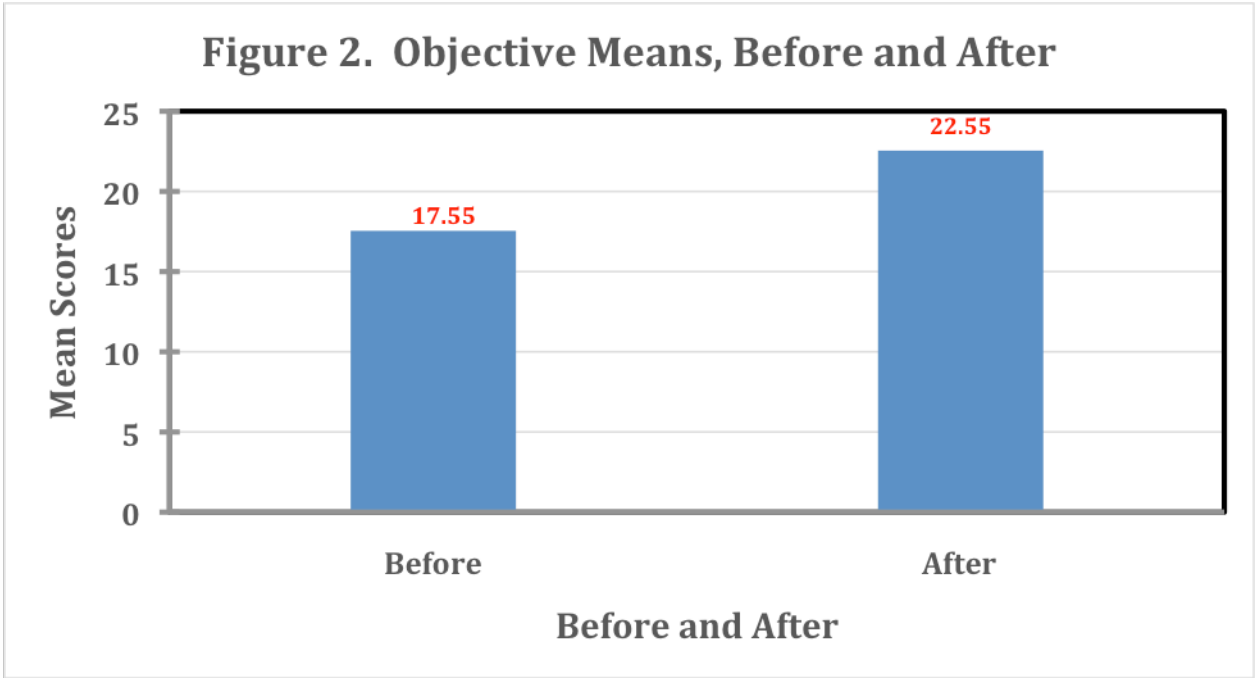


Figure 2. Objectives Means, Before and After

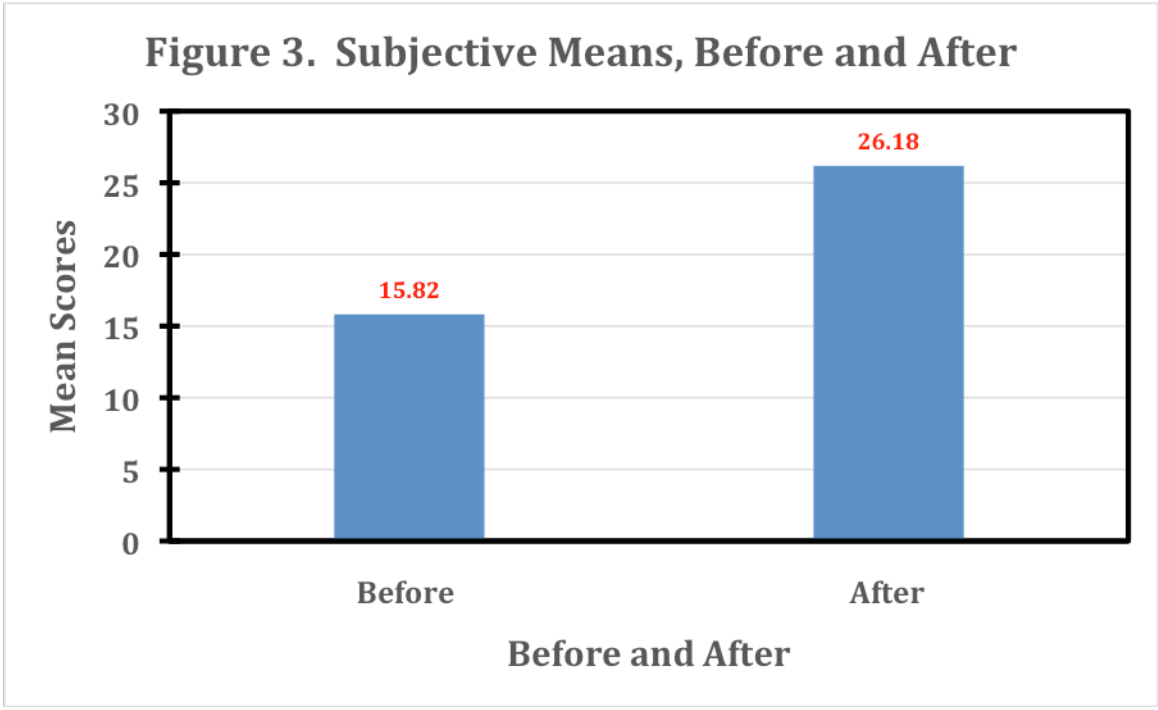


Figure 3. Subjective Means, Before and After

Effect of Years of Experience

The variable "years of experience as a registered nurse" is evaluated both as an effect on initial scores and an improvement in scores. The questionnaire lists intervals for the years of experience. For purposes of the analysis, these are then converted into mid-points as listed in Table 4. The open-ended range of five years and more is converted to seven years.

Table 4. Conversion of Years.

Survey	Mid-point
Less than 1	0.5
1 to 2	1.5
3 to 5	4.0
More than 5	7.0

Linear regressions are done for RN experience against the objective and subjective initial scores and improvement. The intercept and slope define the linear regression equation. y = Intercept + Slope*x, where y is the score and x is the number of years of RN experience. For example, for the first line, y = 14.93 + 0.554*x, where y is the initial score and x is the number of years of experience as an RN. A zero slope indicates that the years of experience has no effect. We are testing that the true slope is zero. A p-value below 0.05 indicates that we should reject the hypothesis of zero slope (that is, no effect). All p-value exceed 0.05. This means that we cannot find a statistical effect due to RN experience.

Table 5. RN Experience Regression

	Intercept	Slope	t	p
Objective Initial	14.93	0.554	1.65	0.115
Objective Improvement	6.68	-0.354	-0.98	0.339
Subjective Initial	17.10	-0.271	-0.87	0.393
Subjective Improvement	9.41	0.202	0.38	0.707

The same regressions were done by the years of experience in pediatrics oncology. While we find no statistical significance in any of the regressions, three of the four regression had low p-values slightly greater than 0.05. This suggests that the relatively small sample size of 22 students could be masking the real effect.

From the signs of the slopes, it appears that more experience leads to higher initial objective scores and then smaller objective score improvements. Similarly, more experience leads to smaller improvements in the subjective scores.

Table 6. Pediatric Oncology Experience Regressions

	Intercept	Slope	t	p
Objective Initial	14.92	0.634	1.85	0.079
Objective Improvement	7.63	-0.635	-1.78	0.090
Subjective Initial	16.30	-0.117	-0.36	0.723
Subjective Improvement	14.66	-1.039	-2.08	0.051

Figures 4, 5, and 6 show plots of the three most clear-cut regressions. The blue dots are the observed data and the red line is the fitted regression line.

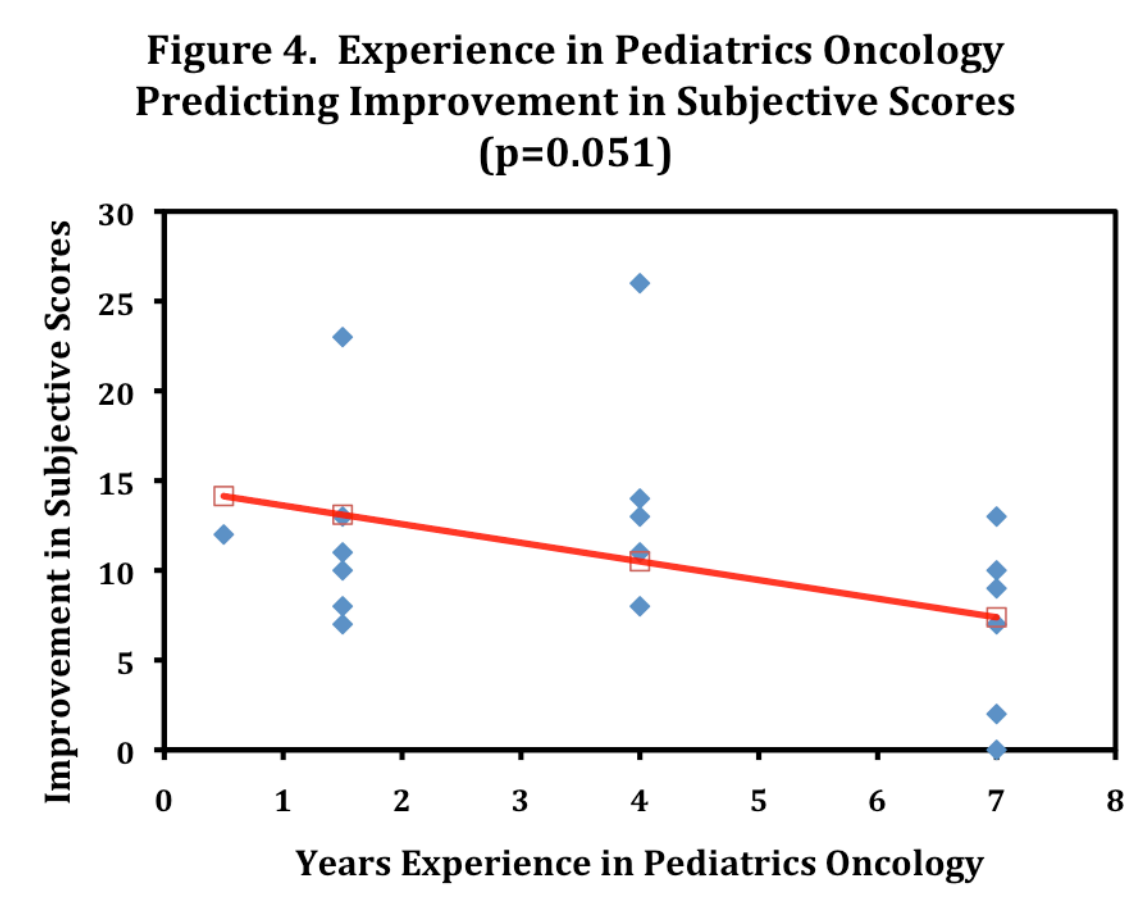


Figure 4. Experience in Pediatric Oncology Predicting Improvement in Subjective Scores

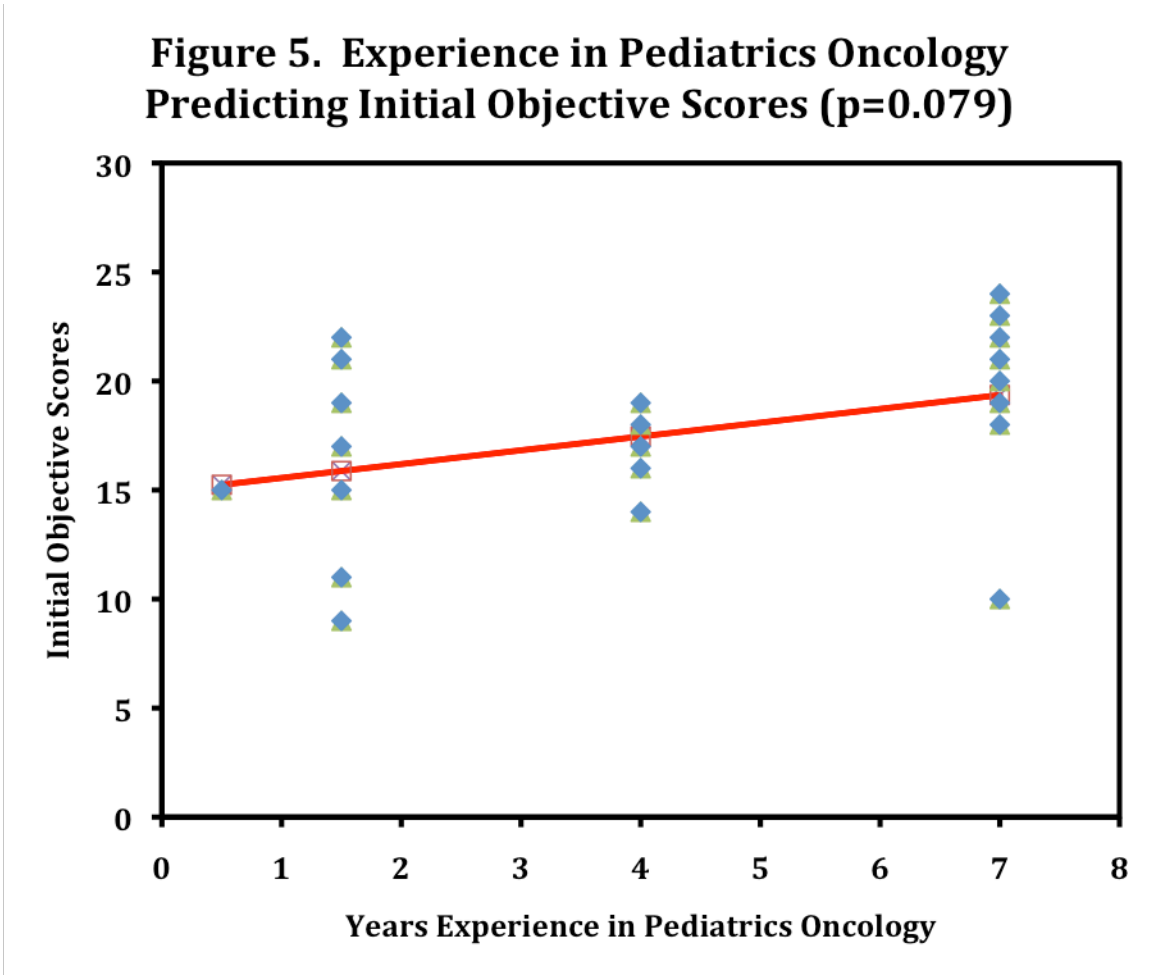


Figure 5. Experience in Pediatrics Oncology Predicting Initial Objective Scores

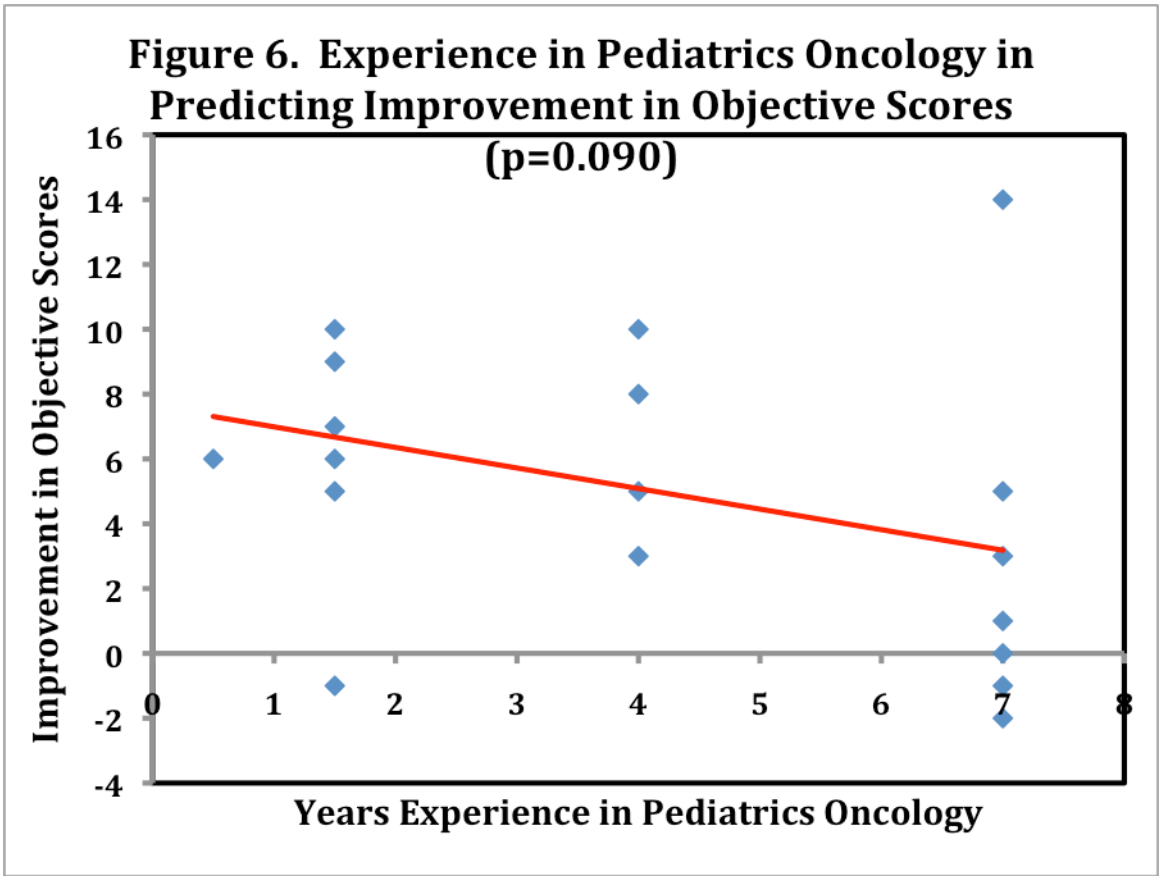


Figure 6. Experience in Pediatrics Oncology in Predicting Improvement in Objective Scores

Effect of Prior NF 1 Experience

We next examine the effect of prior NF 1 education. The five levels are coded 1 through 5 as described in Table 7.

Table 7. Coding NF1 Education

Level	NF 1 Education
1	No education
2	Learned in school, but can't remember
3	Learned in neurology class
4	Learned in genetics class
5	Learned from experience

The improvement in objective scores is summarized by the mean improvements for each of the five levels in table 8.

Table 8. NF1 Education versus Improvement in Objective Scores

NF 1 Education	Means	Count
1	3.25	8
2	3.83	6
3	5.50	2
4	7.67	3
5	9.00	3

A one-factor analysis of variance (ANOVA) was performed to test if the true improvement means are all equal. The p-value of 0.222 indicates that we cannot reject this hypothesis. Thus, we find no effect due to NF 1 education level.

Table 9. Analysis of Variance for NF1 Education versus Improvement in Objective Scores.

Source of Variation	*SS*	*df*	*MS*	*F*	*p*
Between Groups	102.5	4	25.63	1.59	0.222
Within Groups	273.5	17	16.09		
Total	376	21			

Tables 10 and 11 list the analogous results for the improvement in subjective scores. Again, we find no statistically significant effects due to prior NF 1 education level.

Table 10. NF1 Education versus Improvement in Subjective Scores

NF 1 Education	Means	Count
1	11.29	8
2	5.00	6
3	8.00	2
4	11.50	3
5	13.50	3

Table 11. Analysis of Variance for NF1 Education versus Improvement in Subjective Scores

Source of Variation	*SS*	*df*	*MS*	*F*	*p*
Between Groups	165.8	4	41.45	1.50	0.264
Within Groups	332.4	17	27.70		
Total	498.2	21			

Correlations Between Objective and Subjective Scores

A correlation coefficient between two variables is a number between -1 and +1. A correlation of +1 indicates perfect correlation, meaning that if you know one of the variables, you know the other. A correlation of 0 means that the two variables are unrelated. A correlation of -1 means that they are perfectly related, but go in opposite directions. We look at the correlation between the objective and subjective scores, with respect to the initial scores as well as the improvement. These correlations are summarized in Table 12. The p-values indicate that these correlations are not statistically significant.

Table 12: Correlations between Objective and Subjective Test Scores

Pairs	Correlation	p
Initial Objective and Subjective	0.101	0.656
Improvement Objective and Subjective	0.287	0.195

CHAPTER 5: CONCLUSION

Discussion

The results from the pre-test and post-test surveys revealed information about several aspects of nursing NF1 knowledge and education. When asked about former NF1 education, the majority of nurses claimed to have no education (36.36%). Twenty-seven percent of the participants stated to have heard about NF1 in nursing school but did not remember anything, and 18.18% said they only learned about NF1 from prior experience working with NF1 patients. The smallest percentages of nurses remembered learning about this population in either neurology lectures (9.09%) or genetics lectures (9.09%). When considering the prevalence of pediatric NF1 and the likelihood of encountering an NF1 patient while working as a nurse, these reports of NF1 education are surprising. This may be explained by the length of time since graduating from an academic nursing institution seeing as how 50% of the subjects had been a nurse for >5 years and may not remember former NF1 education. Additionally, the majority of participants had worked specifically in a pediatric oncology department for >5 years (36.36%) and may not utilize NF1 knowledge regularly. While the AACN's *Essentials of Baccalaureate Education for Professional Nursing Practice* mentions the responsibility of nursing baccalaureate programs to prepare graduates to be able to assess predictive factors such as genetics, academic programs may neglect to cover certain content (AACN, 2008). With so many genetic disorders to include in undergraduate nursing curricula, nursing instructors may not be choosing to discuss NF1 directly. Pre-test results indicate that approximately one third (36.36%) of nurses responded "definitely not" when asked how prepared they feel to adequately care for pediatric NF1 patients, one

third (31.82%) chose "not really" and one third (31.82%) "yes, but I'd like more education." None of the subjects responded "Yes, I feel confident." With a generalized need to equip nurses with genomic and genetic competencies, it is particularly indicated within a setting like the infusion center at which NF1 patients will receive care.

Results also helped determine the effectiveness of the NF1 educational tool (Power Point). In efforts to meet the nursing NF1 education project outcome of creating an effective nursing educational tool, responses from the pre-test were compared to those from the post-test to determine if, indeed, the educational tool improved nursing NF1 knowledge. The mean objective score, as well as the mean subjective score, both improved from pre-test (17.55) to post-test (22.55) with statistical significance, suggesting that nurses gained NF1 knowledge by reviewing the NF1 educational tool. The subjective mean pre-test score (15.82) significantly increased with the post-test (26.18) as well, telling us that the educational tool succeeded in effectively improving nursing NF1 knowledge perceptions and confidence. The project's goal of creating a NF1 educational tool that demonstrated effective improvements in nursing NF1 knowledge and nursing NF1 knowledge perceptions was met.

Interestingly, the number of years with registered nursing (RN) experience nor the amount of prior experience working with NF1 patients had a significant effect on mean objective or mean subjective scores. The years of RN experience or NF1 experience did not affect the amount of improvement in scores between the pre- and post-tests. However, the years experience working with pediatric oncology were related to the improvement in subjective scores, as well the objective initial and objective improvement scores. The more years of experience working with pediatric oncology patients resulted in higher objective scores and

then smaller improvements. These were all not quite significant at the 5% significance level, but were close. It may be that the relatively small sample size made it difficult for a real effect to be formally detected. It is unclear the reason for this near-significant statistical finding. Possibly nurses with oncology backgrounds have exposure to additional training or education regarding genetics. Cancer nursing courses have been used as platforms to introduce nursing genomics and genetics, based off the fact that cancer is essentially inseparable from the genetics concepts (Kiernan & Vallerand, 2016). Cancer education has become a mainstream topic in undergraduate nursing curricula and therefore incorporating genes and gene expression into the standard malignancy lectures is logical. Oncology courses have been used as a way to meet genetic education requirements stated in the AACN nursing baccalaureate guidelines (Kiernan & Vallerand, 2016; AACN, 2008). While specific NF1 information may not have been reviewed in previous academic settings, nurses with greater amounts of exposure to the oncology profession may be applying genetic nursing knowledge to the NF1 population.

The correlation between nursing NF1 knowledge and nursing NF1 knowledge perceptions were found to be insignificant, both in overall scores (correlation = 0.101, p=0.656) as well as in score improvement levels (correlation = 0.287, p=0.195). This suggests that nursing NF1 intelligence does not relate to nursing NF1 confidence, comfort, or sense of preparedness. It is reassuring that when asked "Do you feel prepared to adequately care for the NF1 patients in the infusion center?," the majority of pre-test responses (36.36%) claimed "definitely not" while the majority of post-test responses (52.27%) chose "yes, but I'd like more education." By having a larger sample size, we may have been able to statistically demonstrate that nurses acquired more confidence to care for NF1

patients after having completed the NF1 educational tool. The same phenomena occurred with nursing perceptions of their understanding of the nursing role in NF1 care. 81.82% of nurses on the pre-test stated "poor," 18.18% chose "good" and 0% selected "excellent." On the post-test, 45.45% "poor," 47.43% "good" and 6.82% "excellent." While the nurses' perception of the nursing role improved after utilizing the educational tool, the difference was not significant to demonstrate actual change. This reinforces the importance of a larger sample size.

Limitations

While the nursing NF1 education project was able to contribute to the knowledge base of infusion center nurses as they prepare to care for pediatric NF1 patients, limitations are present in this study. The most considerable limitation is the study's small sample size, which prohibited the statistical analysis from concluding information about the effectiveness of the NF1 educational tool, nursing NF1 knowledge, and nursing NF1 knowledge perceptions. It is not clear if nurses were motivated to participate in this survey for there was no compensation involved. In addition, the subjects were from one geographical location, and from the same department all within the same hospital. This hinders the ability to generalize the information collected in this project to other nursing departments across California and in other states. Additionally, there was no pilot study conducted to test the validity or difficulty level of the test questions. It would have been helpful to initially administer the pre-test, educational tool, and post-test to a small sample of non-nurses and nurses. The results from this pilot study would help determine whether the test questions were too easy or difficult, and how scores compare between the general public and the nursing workforce. This information could then gauge the test questions that were ultimately used for

the pre-test and post-test within this project. Another limitation was the lack of post-test questions that related to the nurses' opinions of the educational tool directly. Information regarding how well they navigated through the educational tool PowerPoint, if they had any unanswered questions about the presented information, or their general opinion about the electronic self-guided PowerPoint learning method, would have provided insight into this teaching style. Lastly, one more limitation to this project was the inability to compare the effectiveness of this NF1 educational tool to another pre-existing tool. In Munroe and Loerzel's 2016 study that assessed nursing student's knowledge of genomic concepts by using the Genomic Nursing Concept Inventory (GNCI), authors were able to compare their results (45% on pre-test to 50% on post-test) to a previous study that measured knowledge gain from the GNCI in 2013 (44% on pre-test to 79% on post-test), (Munroe & Loerzel, 2016). Because there exists no other educational tool to measure nursing NF1 knowledge, a similar comparison was unattainable.

Conclusion

In summary, nurses are expected to perform essential genetic and genomic competencies and be equipped to provide safe and appropriate care for all patients with genomic and genetic conditions (ANA, 2009; AACN, 2008). Despite this mandate, it is difficult to find examples in the literature that demonstrate these competencies in action within the clinical setting (Kiernan & Vallerand, 2016). NF1 is a genetic disorder affecting approximately 1 in 3,000 people and requires a multidisciplinary team of medical professionals to meet the recommended management guidelines (CTF, 2016). There is a likelihood that pediatric nurses will, at some point in their careers, encounter the opportunity to care for an NF1 patient. With the use of an effective nursing NF1 educational tool, nurses can

familiarize themselves with the disorder, gain confidence in providing care to this population, and ultimately meet the genomic and genetic competencies established by the ANA and the AACN.

This project set forth to create such an educational tool whereas one currently does not exist, hoping to achieve three outcomes: (1) produce an effective educational tool that improved nursing NF1 knowledge and nursing NF1 knowledge perceptions, (2) support the mission of the Children's Tumor Foundation to drive research, expand knowledge, and advance care for the NF community, and (3) contribute to the educational objectives of the AACN by achieving the essential nursing competencies expected of all nursing graduates. While an educational tool was created that was shown to be significantly effective in improving nursing NF1 knowledge and NF1 knowledge perceptions, a larger sample size is needed to determine correlation and relevance to previous NF1 education, nursing experience, and overall comfort in caring for these patients. Exposing nurses to the educational tool, however, contributes to the academic efforts of enhancing nursing awareness of the NF1 population and medical needs, fulfilling the intentions of the CTF. The educational tool also aligns with AACN's foundational outcome competencies deemed essential for all nurse baccalaureate graduates, regardless of specialty or focus (AACN, 2008). This document mentions genomics or genetics skill sets in 4 areas. Within Essential I, nurses are expected to endure a liberal education involving science, the arts, and life science such as biology and genetics. Within Essential V, nurses are expected to understand a broader context of health care as it impacts social trends, such as within the ever changing science of genomics and genetics. Within Essential VII, it states that the "baccalaureate program is expected to prepare the student to assess protective and predictive factors, including genetics, that influence health of

individuals, families, communities, and populations," (AACN, 2008, pg 24). And within Essential IX, the AACN states that both genetics and genomics are fields where knowledge is constantly expanding. Nurses should be cognizant of tailored therapies designed to improve patient care outcomes," (AACN, 2008, pg 30). Sharing the same educational goals as these essentials, the nursing NF1 educational tool contributes to the overall mission of improving nursing genetic and genomic knowledge so that nurses are empowered to take part in the health, prevention, screening, treatment selection, treatment effectiveness, and constructed pedigrees from family histories (AACN, 2008).

General themes arising from this project include awareness of the ongoing need for nursing knowledge assessments. It is evident that nurses often learn care techniques on the job or from colleagues. Whether in a structured academic setting or not, nurses should be routinely assessed for confidence and competency with tasks involving patient care. This will determine the need for further education, investigate the use of evidence-based research in everyday practice, and monitor patient safety. In addition, the nursing NF1 education project emphasizes the ongoing need to review the expected nursing competencies outlined by the ANA and AACN. It is critical that both nursing schools and nurse managers remain versed in the expectations of nursing competencies, including those relating to genetic and genomic skills, so that nurses not only receive the expected education, but that it translates to clinical practice. A final theme to acknowledge is one that highlights the importance of listening to patients' needs. Parents with children who have NF1 described feeling frustrated and angry because of experiences with health care professionals who had not heard of NF1 or who misunderstood it (Barke et al., 2016). Members of the medical community should interpret this and recognize the professional responsibility and obligation of

educating themselves in order to help provide comfort and reassurance to these families.

Recommendations for future expansion of this project include testing the effectiveness of this nursing NF1 educational tool to larger sample sizes so as to extract a more meaningful and significant conclusion relating to its ability to improve NF1 knowledge. With stronger evidence to suggest that this tool achieves its goals, it could then be introduced to larger NF1 resource platforms such as the CTF and be incorporated in nursing school curricula. Nurses will be able to reference this tool when seeking standardized NF1 knowledge and nursing guidelines, or potentially earn continuing education credits/units with its use. The NF1 educational tool can ultimately contribute to nursing knowledge, help achieve competencies in alignment with the ANA and the AACN guidelines, and support nurses in providing appropriate care for NF1 patients.

REFERENCES

REFERENCES

American Academy of Pediatrics. (2008). Joseph Hersh, Committee on Genetics. Health supervision for children with neurofibromatosis. *Pediatrics, 121*(3), 633-642. Doi: 10.1542/peds.2007-3364. Retrieved from https://pediatrics.aappublications.org/content/121/3/633.

American Association of Colleges of Nursing. (2008). *The essentials of baccalaureate education for professional nursing practice*. Retrieved from http://www.aacnnursing.org/Portals/42/Publications/BaccEssentials08.pdf

American Association of Colleges of Nursing. (2011). Essential genetic and genomic competencies for nurses with graduate degrees. Retrieved from https://www.aacnnursing.org/Portals/42/AcademicNursing/CurriculumGuidelines/Essenetials-Genetic-Genomic-Competencies-2011.pdf

American Nurses Association. (2009). *The essentials of genetic and genomic nursing: competencies, curricula guidelines, and outcome indicators, 2nd Ed.* [PDF file]. Retrieved from https://www.genome.gov/pages/careers/healthprofessionaleducation/geneticscompetency.pdf

American Nurses Association (2016). Genetics and personalized medicine. Retrieved from https://www.nursingworld.org/practice-policy/nursing-excellence/ethics/genetics/

Barke, J. Coad, J., & Harcourt, D. (2016). Parents' experience of caring for a young person with neurofibromatosis type 1 (NF1): a qualitative study. Journal of Community Genetics, 7(1), 33-398. http://doi. 10.1007/s12687-015-0247-z

Baker, M. (2012). Education requirements for nurses working with people with complex neurological conditions: nurses' perceptions. Nurse Education Today, (32), 71-77. https://doi.org/10.1016/j.nedt.2011.01.011

Boyd, K.P, Korf, B.R., and Theos, A. (2009). Neurofibromatosis type 1. *Journal of the American Academy of Dermatology, 61*(1), 1-14. http://doi.org/10.1016/j.jaad.2008.12.051

Calzone, K.A., Jenkins, J., Culp, S., and Badzek, L. (2018). Hospital nursing leadership-led interventions increased genomic awareness and educational intent in magnet settings. Nursing Outlook, 66, 244-253. http://doi.org/10.1016/j.outlook.2017.10.010

Camak, D.J. (2016). Increasing importance of genetics in nursing. *Nurse Education Today, 44*, 86-91. https://ac-els-cdn-com.hmlproxy.lib.csufresno.edu/S0260691716300764/1-s2.0-S0260691716300764-main.pdf?_tid=190d2332-eb9f-4d41-8f07-27ce55881c59&acdnat=1531709549_d2df54c53d8ba579a8f7598967404c48

Chen, L. and Kim, M. (2014). Needs assessment in genomics education: a survey of health educators in the United States. *Health Promotion Practice, 15*(4), 592-598. Doi: 10.1177/1524839913483470.

Children's Tumor Foundation. (2016). *NF1*. Retrieved from http://www.ctf.org/understanding-nf/nf1.

Clapper, T.C. Beyond Knowles: what those conducting simulation need to know about adult learning theory. Clinical Simulation in Nursing, (6)1, e7-e14. https://doi.org/10.1016/j.ecns.2009.07.003

Connors, L. and Schorn, M. (2018). Genetics and genomic content in nursing education: a national imperative. Journal of Professional Nursing, 34(4), 235-237. https://doi.org/10.1016/j.profnurs.2018.06.003

Draucker, C.B., Nutakki, K., Varni, J.W., and Swigonski, N.L. (2016). The health-related quality of life of children, adolescents, and young adults with neurofibromatosis type 1 and their families: analysis of narratives. Journal for Specialists in Pediatric Nursing, 22. Https://doi.org/10.1111.jspn.12174

Evans, D.G.R., Salvador, H., Chang, V.Y., Erez, Ayelet, Voss, S.D., Schneider, K.W., Scott, H.S., Plon, S.E., & Tabori, U. (2017). Cancer and central nervous system tumor surveillance in pediatric neurofibromatosis 1. Clinical Cancer Research, 23(12), 46-51. Doi: 10.1158/1078-0432.CCR-17-0589

Ferner, R. E., Huson, S. M., Thomas, N., Moss, C., Willshaw, H., Evans, D. G., Upadhyaya, M., Towers, R., Gleeson, M., Steiger, C., and Kirby, A. (2006). Guidelines for the diagnosis and management of individuals with neurofibromatosis 1. *Journal of Medical Genetics*, *44*(2), 81-8. Doi: 10.1136/jmg.2006.045906

Gallo, A.M., Angst, D.B., Knafl, K.A., Twomey, E.H. (2010). Health care professionals' views of sharing information with families who have a child with a genetic condition. Journal of Genetic Counseling, 19(3), 296-304. Https://doi-org.hmlproxy.lib.csufresno.edu/10.1007/s10897-010-9286-0

Genomic Nursing State of the Science Advisory Panel, Calzone, K.A., Jenkins, J., Bakos, A.D., Cashion, A.K., Donaldson, N., Feero, W.G., Feetham, S., Grady, P.A., Hinshaw, A.S., Knebel, A. R., Robinson, N., Ropka, M.E., Seibert, D., Stevens, K.R., Tully, L.A., Webb, J.A. (2013). A blueprint for genomic nursing science. *Journal of nursing scholarship : an official publication of Sigma Theta Tau International Honor Society of Nursing*, *45*(1), 96-104. https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3594405/

Getha-Eby, T.J., Berry, T., Xu, Y., and O'Brian, B.A. (2014). Meaningful learning: theoretical support for concept-based teaching. Journal of Nursing Education, 53(9), 494-500. https://doi-org.hmlproxy.lib.csufresno.edu/10.3928/01484834-20140820-04

Hartzell, J.D. (2007). Adult learning theory in medical education. The American Journal of Medicine, 120(11), e11. http://doi-org.hmlproxy.lib.csufresno.edu/10.1016/j.amj.med.2006.10.024.

Hood, L. and Rowen, L. (2013). The human genome project: big science transforms biology and medicine. *Genome Medicine, 5*(9), 79. Doi: 10.1186/gm483

Institute of Medicine. (2011). The future of nursing: leading change, advancing health. *Committee on the Robert Wood Foundation Initiative on the Future of Nursing, at the Institute of Medicine.* Washington, D.C.: The National Academies Press. ISBN: 987-0-309-15823-7. https://www.ncbi.nlm.nih.gov/books/NBK209885/

Julian, N., Edwards, N. E., DeCrane, S., and Hingtgen, C. M. (2014). Neurofibromatosis 1: diagnosis and management. The Journal for Nurse Practitioners, 10(1), 30-35. https://doi-org.hmlproxy.lib.csufresno.edu/10.1016/j.nurpra.2013.07.001

Kiernan, J. and Vallerand, A.H. (2016). Cancer as a platform for genetics education in the undergraduate nursing curriculum. Journal of Nursing Education, 55(4), 236-239. https://doi-org.hmlproxy.lib.csufresno.edu/10.3928/01484834-20160316-11

Kenner, C. and Weinerman, J. (2011). Adult learning theory: applications to non-traditional college students. Journal of College Reading and Learning, 41(2), 87-96. Retrieved from http://search.ebscohost.comhmlproxy.lib.scufresno.edu/login.aspx?direct=true&db=ehh&AN=60202793&site=ehost-live

Knowles, M.S., Holton, I.E.F., & Swason, R.A. (2005). *The adult learner: the definitive classic in adult education and human resource development.* Retrieved from https://ebookcentral.proquest.com

Lea, D. H. (2009). Basic genetics and genomics: a primer for nurses. *Online Journal of Issues in Nursing, 14*(2), 3. www.nursingworld.org/MainMenuCategories/ANAMarketplace/ANAPeriodicals/OJIN/TableofContents/Vol142009/No2May09/Arti cles-Previous-Topics/Basic-Genetics-and-Genomics.aspx.

Lessic, M. and Anderson, L. (2000). Genetic discoveries: challenges for nurses who care for children and their families. Journal for Specialists in Pediatric Nursing, 5(1), 47-51. https://doi.org/10.1111/j.1744-6155.2000.tb00085.x

Lewis, J. A. , Calzone, K. M. & Jenkins, J. (2006). Essential Nursing Competencies and Curricula Guidelines for Genetics and Genomics. MCN, The American Journal of Maternal/Child Nursing, 31(3), 146-153. Retrieved from https://oce-ovid-com.hmlproxy.lib.csufresno.edu/article/00005721-200605000-00004/HTML

Messner, R. and Smith, M.N. (1986). Neurofibromatosis: relinquishing the masks; a quest for quality of life. *Journal of Advanced Nursing, 11*, 459-464. https://onlinelibrary-wiley-com.hmlproxy.lib.csufresno.edu/doi/epdf/10.1111/j.1365-2648.1986.tb01273.x

Montgomery, S., Brouwer, W.A., Everett, P.C., Hassen, E., Lowe, T., McGreal, S.B., Eggert, J. (2017). Genetics in the clinical setting: what nurses need to know to provide the best patient care. *American Nurse Today, 12*(10), 2017. Retrieved from https://www.americannursetoday.com/genetics-clinical-setting/

Munroe, T. and Loerzel, V. (2016). Assessing nursing students' knowledge of genomic concepts and readiness for use in practice. *Nurse Educator, 41*(2), 86-89. Doi: 10.1097/NNE.0000000000000201.

National Center for Biotechnology Information . (2018). *NF1 neurofibromin 1*. Retrieved from https://www.ncbi.nlm.nih.gov/gene/4763.

National Council of State Boards of Nursing (NCSBN), 2018. NCLEX-RN Examination: Test plan for the national council licensure examination for registered nurses. Retrieved from https://www.ncsbn.org/2019_RN_TestPlan-English.pdf

National Human Genome Research Institute, 2016. An overview of the human genome project. Retrieved from https://www.genome.gov/12011238/an-overview-of-the-human-genome-project/

National Institute of Health. (2018). *Genetics home reference: neurofibromatosis type 1*. Retrieved from https://ghr.nlm.nih.gov/condition/neurofibromatosis-type-1#genes.

National Institute of Health. (2016). *National Human Genome Research Institute: learning about neurofibromatosis*. Retrieved from https://www.genome.gov/14514225/learning-about-neurofibromatosis/.

National Institute of Health (1987). *NIH Consensus Development Program: Office of Disease Prevention*. Retrieved from https://consensus.nih.gov/1987/1987Neurofibramatosis064html.htm

Neurofibromatosis California (n.d.). Retrieved from http://www.nfcalifornia.org/

Neurofibromatosis Network. (2018). *Neurofibromatosis Network Mission*. Retrieved from https://www.nfnetwork.org/about/nf-network-mission/

Rasmussen, S.A. and Friedman, J.M. 2000. NF1 gene and neurofibromatosis 1. American Journal of Epidemiology, 151(1), 33-40.

Sampson, J., Thompson, H.L., and Wall Parilo, D.M. (2019). Caring for children with neurofibromatosis type 1. *Nursing, 49*(1), retrieved from www.nursing2019.com. Doi: 10.1097/01.nurse.0000554214.17051.d9

Thompson, H. and Brooks, M.V. (2011). Genetics and genomics in nursing: evaluating *Essentials* implementation. Nurse Education Today, 31(6), 623-627. Https://doi.org/10.1016/j.nedt.2010.10.02

United States Census Bureau. (n.d.). Retrieved January 5, 2019, from https://www.census.gov/quickfacts/santaclaracountycalifornia

APPENDICES

APPENDIX A: SUBJECT RECRUITING EMAIL

Each subject was sent an initial introductory recruiting email on January 14th, 2019. The email was sent to the entire email distribution list of BCDH nurses, totaling 50 recipients. The email contained an introductory paragraph, as well as instructions to follow if the subject chose to participate. Attached to the email was a word document consent form, as well as a PowerPoint educational tool. The pre-test survey and the post-test survey were accessed using hyperlinks, which were embedded into the body of the email. See below for a copy of the email:

Hello BCDH nurses,
You have been invited to participate in a brief study looking at nursing knowledge of pediatric NF1 patient care. This study is being done as part of my DNP project at UC-Fresno/San Jose. Please follow the steps below. Complete participation in this study will take approximately 30 minutes, and it will be available to you until Sunday Feb. 3, 2019. Your involvement is greatly appreciated.

1) Read NF consent form (attached document)
2) Complete pre-test survey:
   https://stanforduniversity.qualtrics.com/jfe/form/SV_4ZrMkEpNAafiZJX
3) Review educational tool (attached slide presentation):
4) Complete post-test survey:
   https://stanforduniversity.qualtrics.com/jfe/form/SV_4ZrMkEpNAafiZJX

For questions, please contact Samantha Ingerick, NP
singerick@stanfordchildrens.org

APPENDIX B: SUBJECT CONSENT FORM

CALIFORNIA STATE UNIVERSITY
NORTHERN CALIFORNIA CONSORTIUM
DOCTOR OF NURSING PRACTICE

Caring for the Pediatric Neurofibromatosis Type-1 Patient:
Improving Nursing Knowledge Through an Innovative Educational Tool

You are being asked to participate in this research study because you a nurse within the Bass Center Day Hospital at Lucile Packard Children's Hospital, and may be asked to care for children with neurofibromatosis type-1 (NF1). The purpose of this research study is to evaluate nursing knowledge, and perceived nursing knowledge, of pediatric NF1 patient care before and after reviewing an NF1 educational tool (PowerPoint presentation). The responses to this study may help contribute to enhanced nursing NF1 education and hopefully lead to increased competencies when caring for this population. ***Please complete the pre-test survey, then review the PowerPoint educational tool, then complete the post-test survey.*** *The pre- and post-tests are each 21 questions and include Likert-scale, multiple choice, and ranking questions. Your complete participation will be approximately 30 minutes. Your participation is completely voluntary and your answers will be kept entirely confidential. By completing the pre-test and post-test you give consent to participating in this investigational research study. You have 3 weeks to complete all sections; starting from the time you received this email. Thank you for your time, please click the pre-test link below if you wish to continue.*

Consent: I consent to participating in the Pediatric NF1 Nursing Education study described above. I understand that my participation is voluntary, and that my name and identifiers will be kept entirely confidential.

APPENDIX C: PRE-TEST SURVEY/POST-TEST SURVEY

Pre-Test Survey / Post-Test Survey

1) Is this your pre-test or post-test?
   1 = pre-test
   2 = post-test

2) To familiarize yourself with the testing format: Please answer the following two questions before beginning the survey:
   1 = What is the name of the street you grew up on?
   2 = What is the name of your first pet?

3) How would you rate your current knowledge of neurofibromatosis type-1 (NF1)?
   1 = non-existent, I've never heard of NF1
   2 = I've heard of NF1, but I don't know much about it
   3 = I'm familiar with NF1, I know basics about the disease
   4 = Above average knowledge of NF1
   5 = Highly educated on NF1

4) What former NF1 education have you received?
   1 = no education
   2 = I heard about in nursing school, but I don't remember anything
   3 = I remember learning about it in neurology lectures
   4 = I remember learning about it in genetics lectures
   5 = I only learned from experience while working with these patients

5) Please rank the following:

	Poor	Good	Excellent
Your current knowledge of Neurofibromatosis Type-1 (NF1):			
Your current knowledge of caring for pediatric NF1 patients:			
Your confidence in caring for pediatric NF1 patients:			
Your recognition skills of café-au-lait spots:			
Your comfort in talking with a family about their child's genetic disorder:			
Your comfort in talking with a family about coping with their child's NF1 diagnosis			
Your understanding of the nursing role with pediatric NF1 patients:			
Your understanding of why NF1 patients occasionally need chemotherapy			

Your current knowledge of why NF1 patients are referred to ophthalmologists			
Your familiarity with NF1 diagnosis criteria			
Your current knowledge of the importance of pain assessments for pediatric NF1 patients			
Your ability to list 4 interdisciplinary medical teams involved in the care of pediatric NF1 patients			
Your ability to list 4 nursing interventions that can assist in the care of pediatric NF1 patients			

6) Your experience working as a registered nurse:
A. <1 year
B. 1-3 years
C. 4-5 years
D. >5 years

7) Your experience working within the pediatric oncology population:
A. <1 year
B. 1-3 years
C. 4-5 years
D. >5 years

8) Do you currently feel prepared to adequately care for NF1 patients in the BCDH?
1 = No
2 = Kind of
3 = Yes, but I'd like more education
4 = Yes I feel confident

9) The 3 types of neurofibromatosis (NF) are:
A. NF type-1, NF type-2, and NF type-3
B. NF type-1, NF type-2, and schwannomatosis
C. NF, ependymoma, and medulloblastoma
D. neurofibromin type-1, neurofibromatosis type-1, and neurofibroma type-2

10) For a clinical NF1 diagnosis, a patient must have:
A. six or more café-au-lait macules and freckling in axillary or inguinal regions
B. two or more neurofibromas or one plexiform neurofibroma
C. two or more List nodules
D. optic glioma

E. osseous lesion
F. first-degree relative with known NF1
G. two or more of the symptoms listed above

11) NF1 is a genetic disease that occurs in patients as:
A. an autosomal dominant mutation in the *NF1* gene
B. an autosomal recessive mutation in the *NF1* gene
C. a spontaneous gene mutation
D. A or C
E. A and C

12) Which of the following isn't a potential presentation of NF1?
A. headaches
B. neutropenia
C. learning disabilities/ADHD
D. precocious puberty
E. seizures
F. pain

13) Care for the NF1 patient can involve:
A. ophthalmologists, dermatologists, oncologists and neurologists
B. genetic medicine
C. orthopedic surgery
D. oncologists and cardiologists
E. all of the above

14) Patients with NF1 sometimes develop tumors (neurofibromas) that sometimes cover nerves in the body. These tumors sometimes require the following treatment:
A. bone marrow transplant
B. chemotherapy, surgery, or surveillance scans
C. splenectomy
D. IVIG treatment with scans
E. chemotherapy and splenectomy

15) Individuals affected with NF1 often report __ as major issues impacting their lives:
A. stigma
B. loss of social role and social relationships
C. loss of physical attractiveness and normal body functions
D. decreased educational and financial opportunities
E. all of the above

16) Patients and families affected by NF1 reported that the most frustrating aspect of their health care was:

A. inexperienced health care professions who were unfamiliar with NF1
B. too many social and resources available
C. counting their café-au-lait spots
D. getting MRI scans

17) Which of the following are possible symptoms of NF1 (choose all that apply)

A. Visual impairment (optic glioma)
B. Head: macrocephaly, seizures, brain tumors, learning disabilities
C. Cardio: high blood pressure
D. Liver: elevated ALT and AST liver enzymes
E. Skin: axillary and inguinal freckling, café-au-lait spots
F. Bones: pseudoarthrosis, bone deformities, scoliosis
F. Tumors: neurofibromas that may occur along the nerves
E. Digestive tract: stomach pain, constipation, vomiting

18) As a nurse caring for pediatric NF1 patients, it is important to:

A. Inquire about skin changes, headaches, and changes in vision
B. Avoid touching café-au-lait spots for risk of contagion
C. Measure head circumference of all pediatric NF1 patients
D. Know that all patients will present with identical NF1 symptoms
E. All of the above
F. A & C
G. A & D

19) The pediatric NF1 nursing role includes:

A. Administering chemotherapy to all NF1 patients
B. Identifying and acknowledging any coping difficulties the family might have relating to NF1 diagnosis
C. Avoiding the use of pain scales
D. Recommending that all pediatric NF1 patients receive specialized education for ADHD

20) Clinical changes that could be associated with NF1 presentation include:

A. Recently painful "lump" underneath the skin
B. Recent changes in vision
C. Misalignment of the hips or scoliosis
D. Recent onset headaches
E. Difficulty focusing in school
F. Fever

G. Elevated Blood Pressure

21) All pediatric NF1 patients will present with:

A. List nodules

B. café-au-lait spots

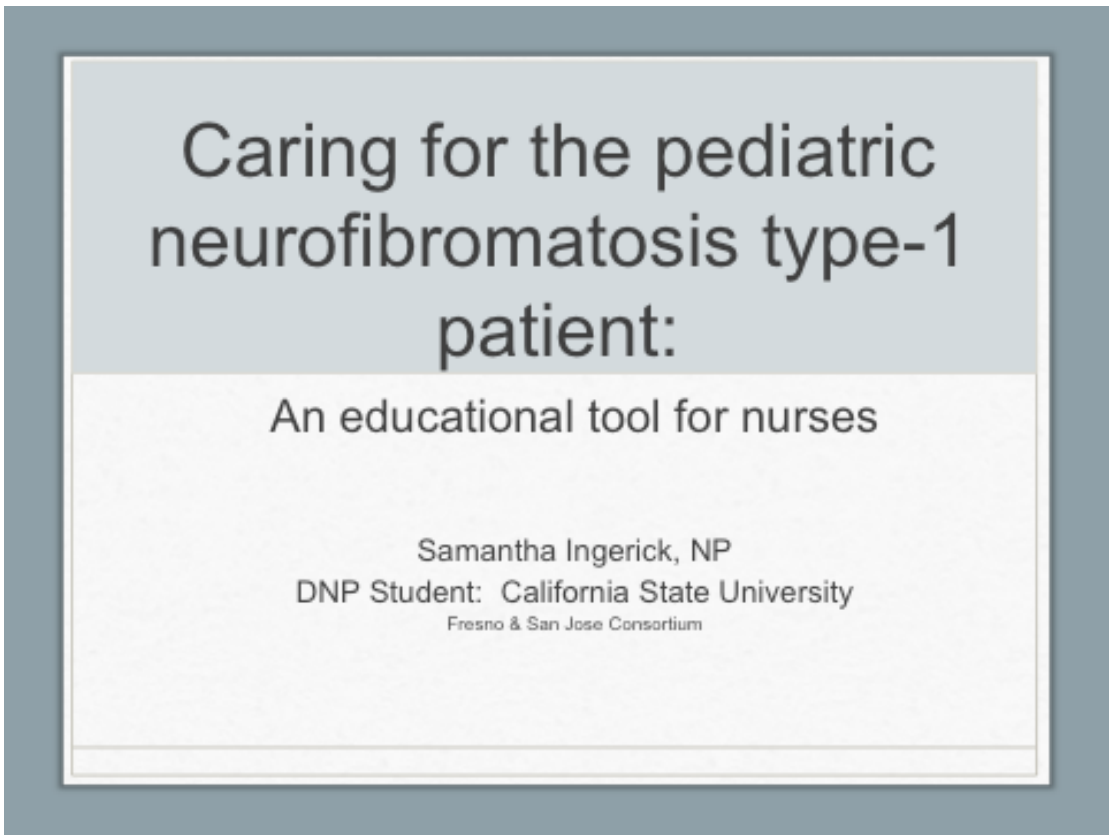
C. a segmental or germ line mutation

D. ADHD

E. tumors

F. all of the above

APPENDIX D: EDUCATIONAL TOOL



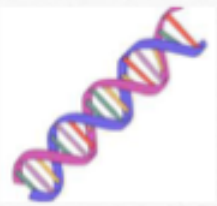
Introduction:

- "Parents describe frustrating situations where health professionals had not heard of NF1, or misunderstood it…. Experiences like this left parents feeling frustrated or angry, or that health professionals would not understand the situation they were in."

Barke, Coad, & Harcourt, 2016

Neurofibromatosis

Neurofibromatosis (NF) is a genetic disorder that causes tumors to grow on nerves throughout the body. *Currently there is no known cure for NF.*



THERE ARE 3 TYPES OF NF:

1) **Neurofibromatosis Type-1 (NF-1):**
   Mutation on gene-17, affects 1 in 3,000 people

2) **Neurofibromatosis Type-2 (NF-2):**
   Mutation on gene-22, affects 1 in 40,000 people

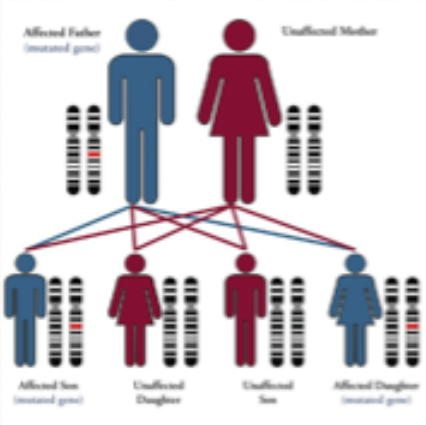
3) **Schwannomatosis:**
   Mutation unidentified, affects less than 1 in 40,000 people

(CTF, 2016)

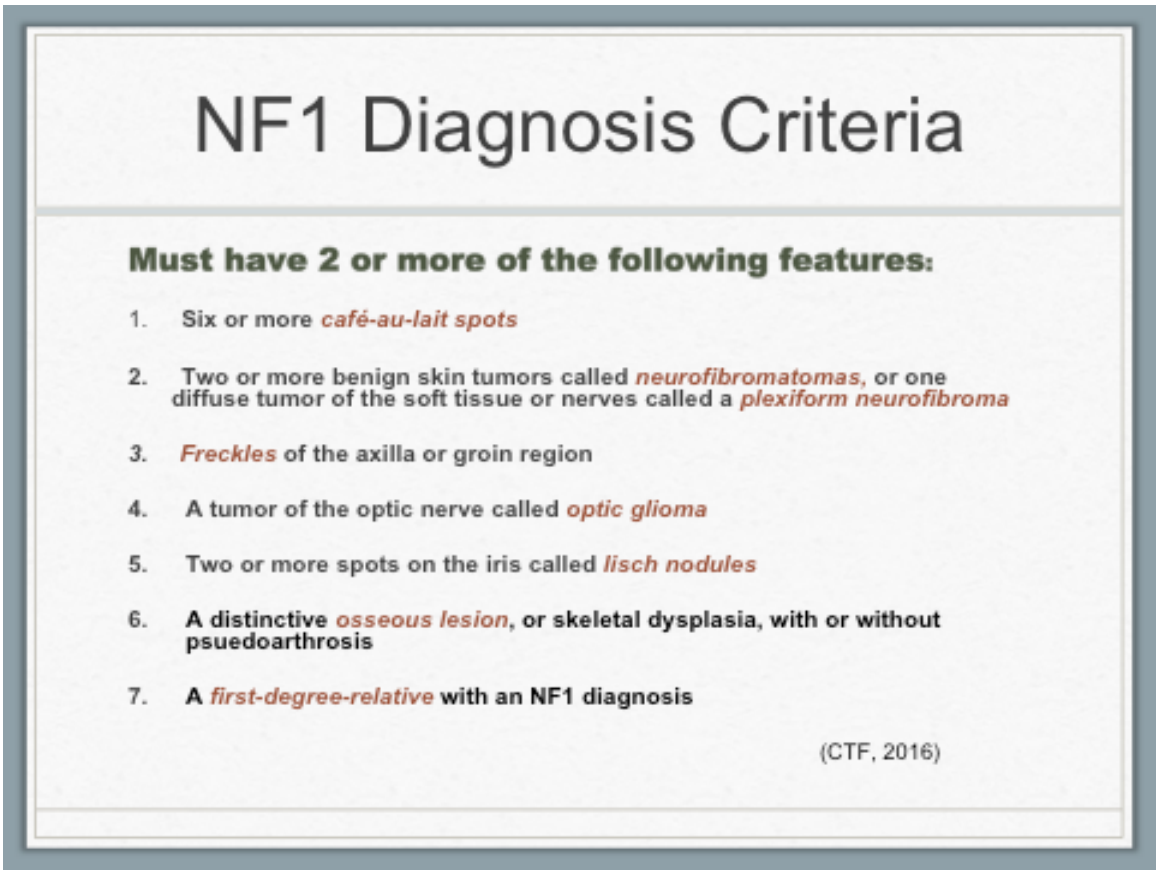
Neurofibromatosis Type-1 (NF1)

- Also known as Recklinghausen's disease
- NF1 is the most prevalent type of NF
- Present at birth
- Either inherited from Parent (autosomal dominant) or spontaneous mutation

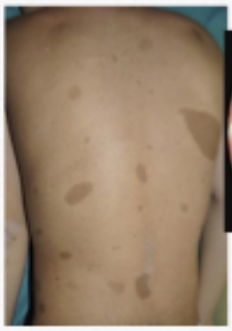
Video



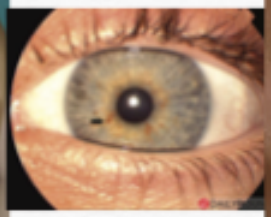
(CTF, 2016)



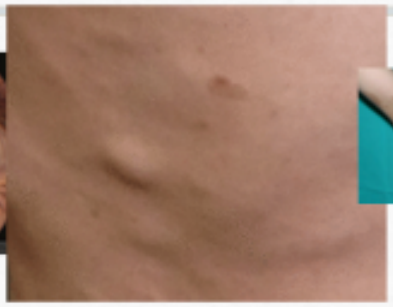
NF1 Diagnosis Criteria



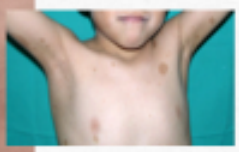
Café-au-lait spots



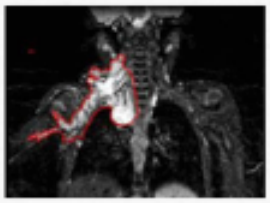
Lisch Nodules



Neurofibroma

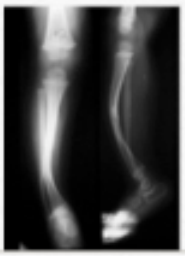


Axillary Freckling



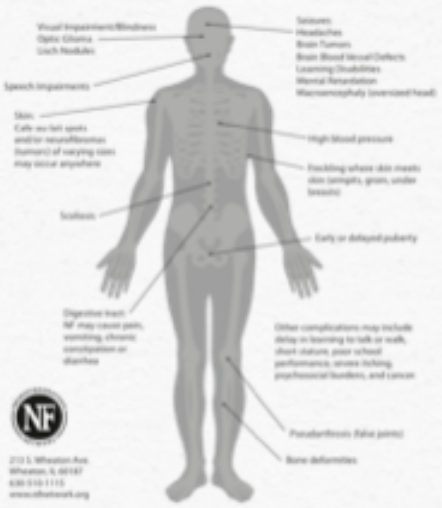
Plexiform Neurofibroma

(Google Images, 2018)



Osseous Lesion

Other NF1 Presentations



Brain	Brain spots on MRI Brain tumors (gliomas)
Skin	Cafe-au-lait spots
Height	Short stature/scoliosis
Eyes	Optic gliomas
Cardio	High blood pressure
Bones	Bone deformities Pseudoarthrosis
GI	Chronic constipation Pain
ADHD	Delayed or early puberty Learning challenges
Puberty	

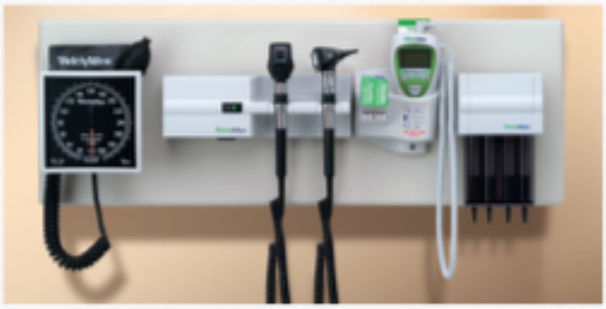
NF1 Management

- NF1 affects ***multiple organs*** and systems
- Most NF1 treatment facilities are adapting the ***medical home model*** from which all care is coordinated between subspecialties
- Longitudinal care is aimed at ***early detection and symptomatic treatment*** as complications occur

NF1 Management

COMPLEX CARE TEAM: There are several variations in clinical NF1 presentations. There may be different specialties caring for each patient:

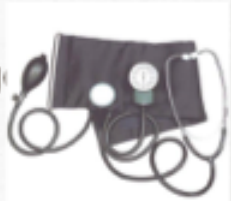
- dermatologist
- ophthalmologist
- geneticist
- orthopedist
- psychologist
- oncologist
- neurologist
- genetic counselor
- school counselor (IEP / 504)



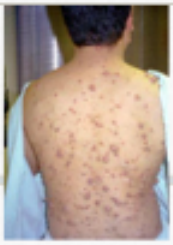
NF1 Management

ROUTINE CARE

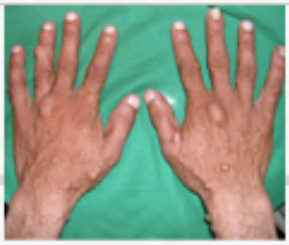
Regardless of the complexity of the NF1 presentation, each patient should undergo ongoing assessments that include:



- **Skin check:** evaluate for new neurofibromas
- **Head circumference:** measure at each visit
- **Blood pressure**: routine checks and PRN. Hypertension may be secondary to renal artery stenosis or aortic stenosis
- **Ophthalmologic evaluations:** annual checks and PRN
- **Skeletal changes:** scoliosis, limb abnormalities
- **Pain:** review pain management plan (AAP, 2008)



Tumors



Neurofibromas (subcutaneous) and plexiform neurofibromas (deeper tissue) may present along the nerves in the body. *These are often not cancerous*, but may cause problems depending on location. These can cause pain, tenderness, and itching. A small percentage of tumors will become malignant.

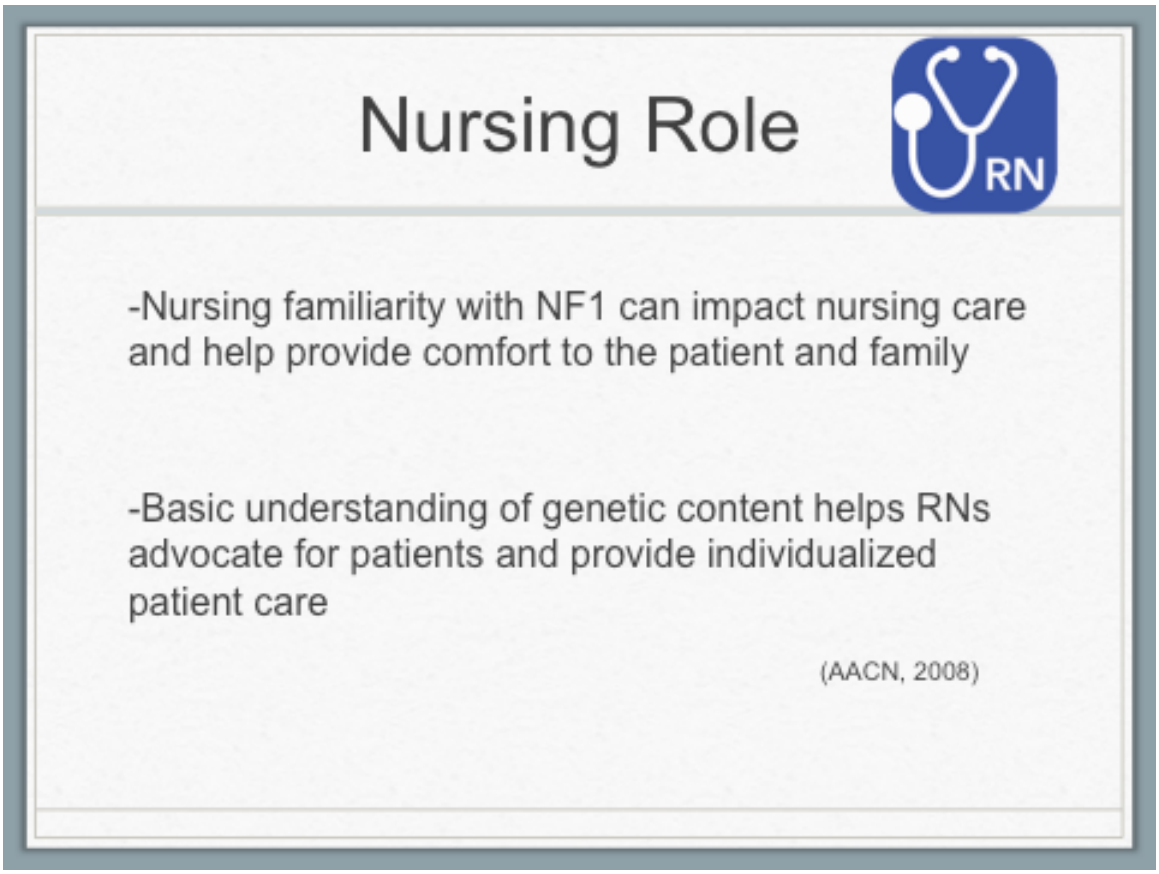
- **Plexiform neurofibromas (PNs) = 25%** (CTF, 2016)
- **Optic pathway gliomas (15%)** (CTF, 2016)
- **Malignant peripheral nerve sheath tumor (2-5%)** (Ferner & Gutman, 2002)

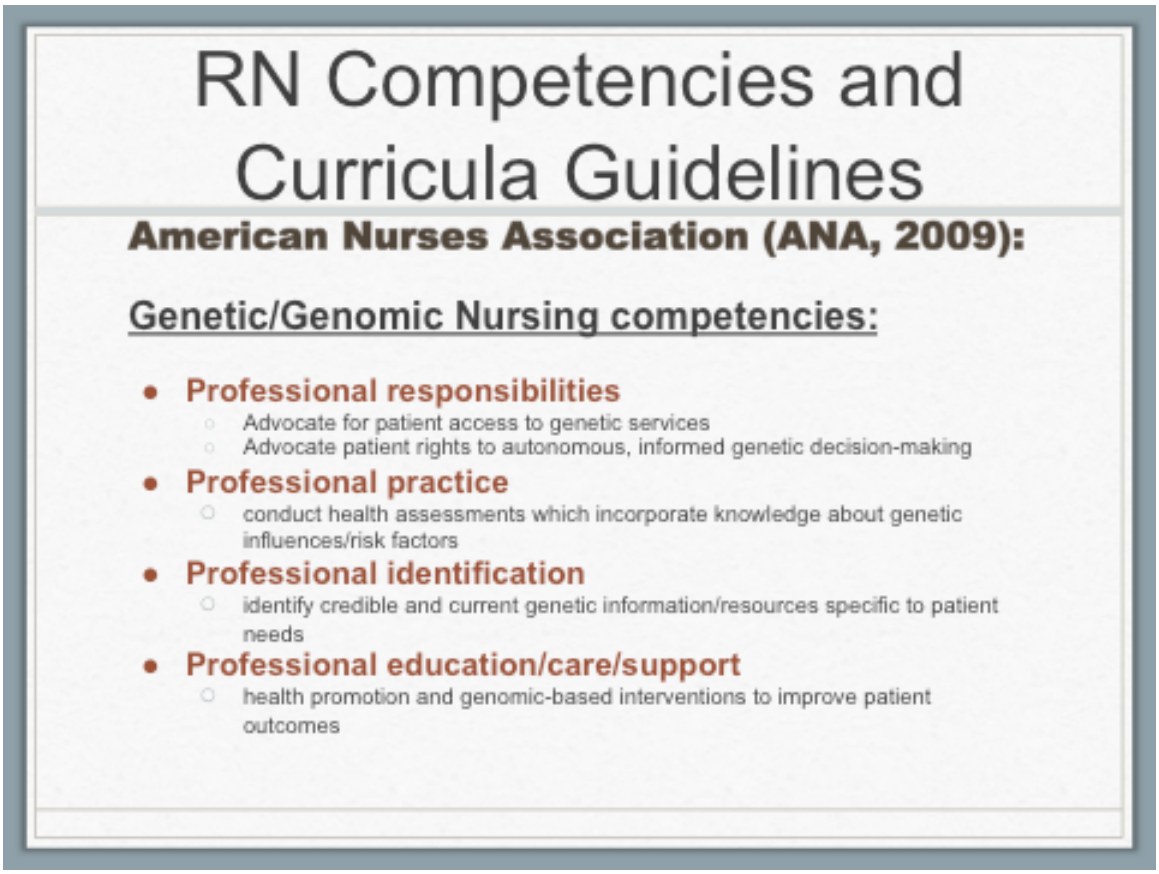
Tumors / Treatment

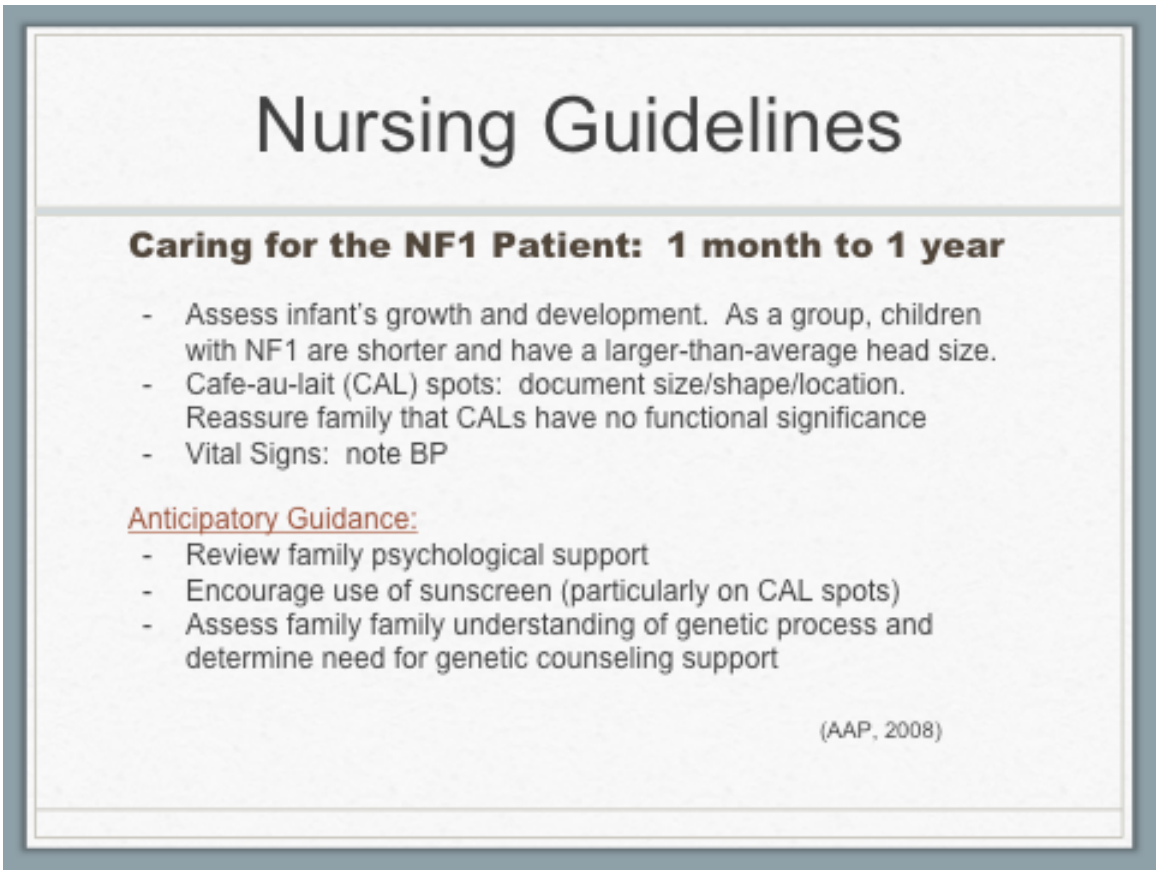
- Currently no cure for NF1 tumors

- **Treatments can help slow tumor growth**
  - **Routine surveillance and MRI scans**
  - **Surgery (tumor debulking)**
  - **Chemotherapy**
    Optic gliomas = carboplatin, vincristine, or other chemotherapies
    Plexiform neurofibromas = oral MEK inhibitors

- Oncology RNs have opportunity to care for these patients during treatments







Nursing Guidelines

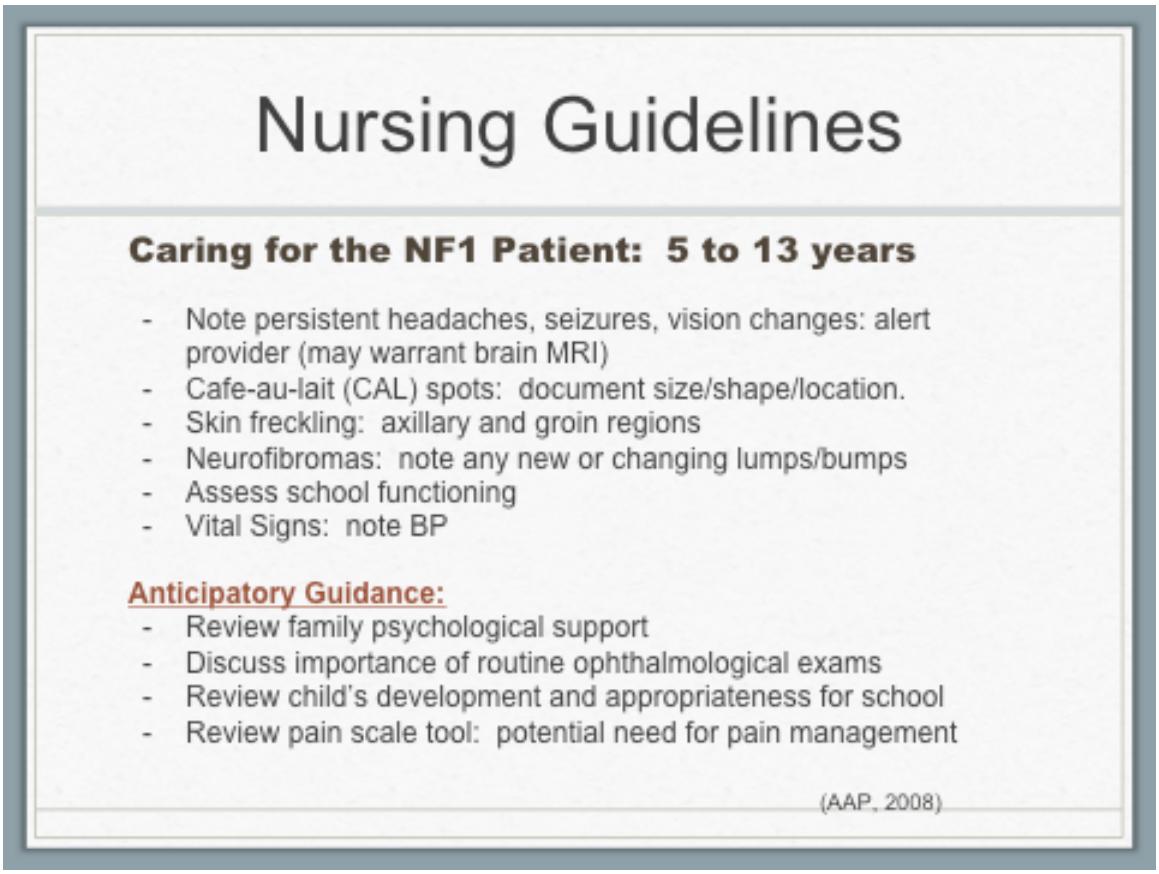
Caring for the NF1 Patient: 1 to 5 years

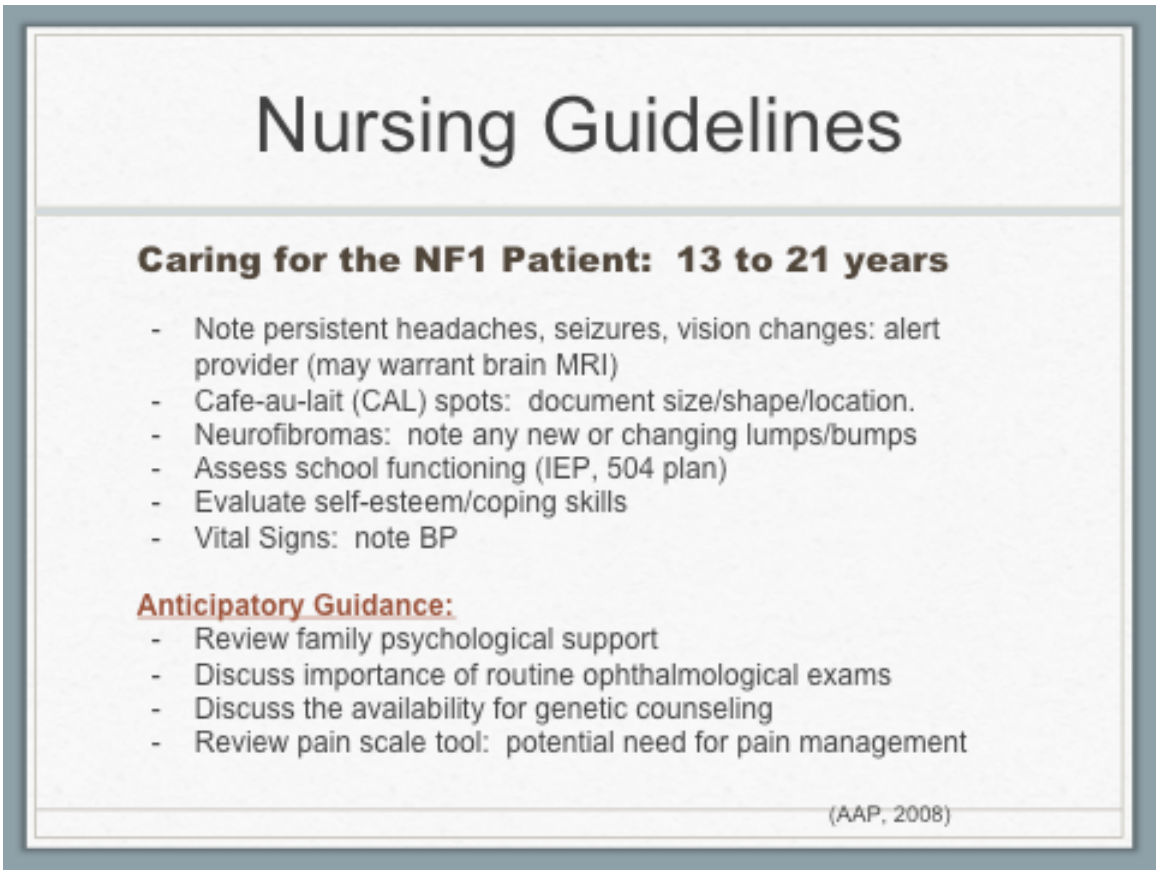
- Note persistent headaches, seizures, vision changes: alert provider (may warrant brain MRI)
- Cafe-au-lait (CAL) spots: document size/shape/location.
- Skin freckling: axillary and groin regions
- Neurofibromas: note any new or changing lumps/bumps
- Vital Signs: note BP

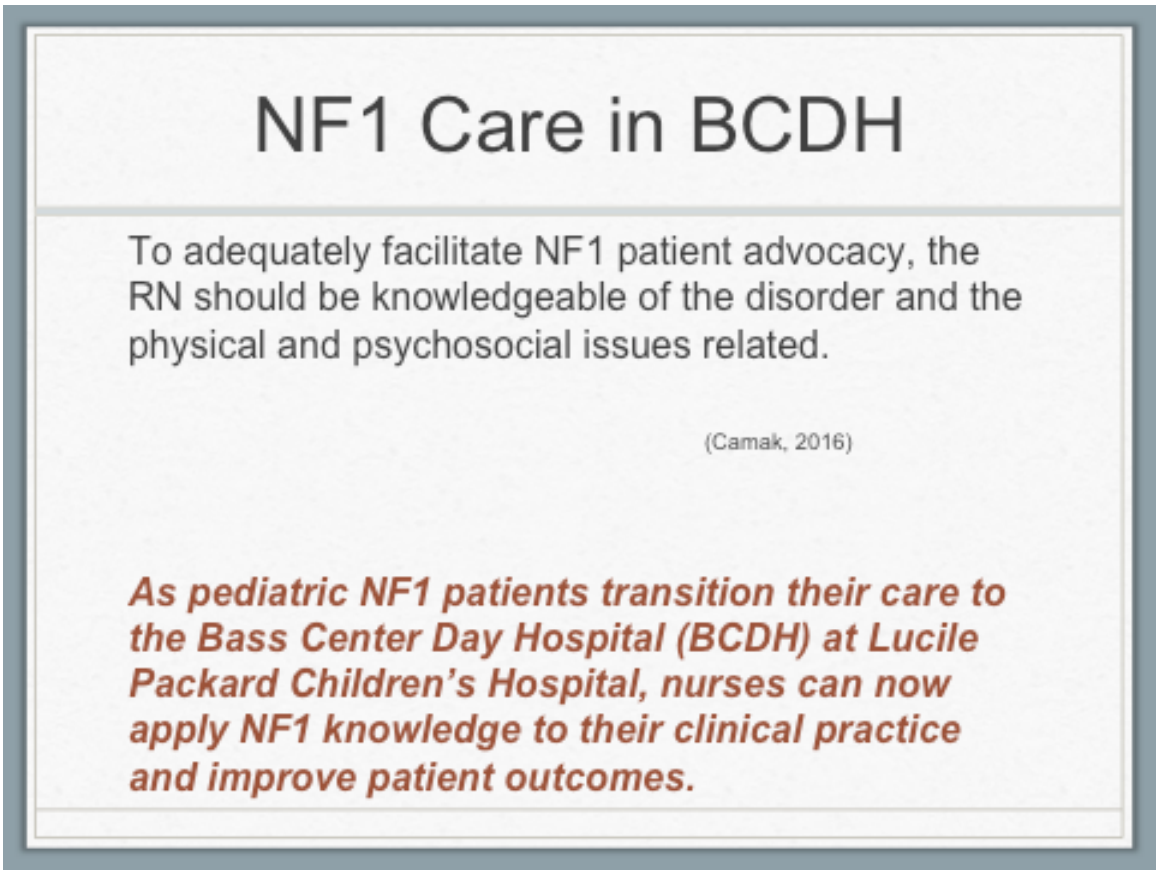
Anticipatory Guidance:

- Review family psychological support
- Discuss importance of routine ophthalmological exams
- Review child's development and appropriateness for school
- Review pain scale tool: potential need for pain management

(AAP, 2008)







References

- American Academy of Pediatrics. (2008). Joseph Hersh, Committee on Genetics. Health supervision for children with neurofibromatosis. *Pediatrics, 121*(3), 633-642. Doi: 10.1542/peds. 2007-3364. Retrieved from https://pediatrics.aappublications.org/content/121/3/633.
- American Association of Colleges of Nursing. (2008). *The essentials of baccalaureate education for professional nursing practice.* Retrieved from http://www.aacnnursing.org/Portals/42/Publications/ BaccEssentials08.pdf
- American Nurses Association. (2009). *The essentials of genetic and genomic nursing: competencies, curricula guidelines, and outcome indicators, 2nd Ed.* [PDF file]. Retrieved from https://www.genome.gov/pages/careers/healthprofessionaleducation/geneticscompetency.pdf
- Barke, J. Coad, J., & Harcourt, D. (2016). Parents' experience of caring for a young person with neurofibromatosis type 1 (NF1): a qualitative study. Journal of Community Genetics, 7(1), 33-398. http://doi. 10.1007/s12687-015-0247-z
- Camak, D.J. (2016). Increasing importance of genetics in nursing. *Nurse Education Today, 44*, 86-91. https://ac-els-cdn-com.hmlproxy.lib.csufresno.edu/S0260691716300764/1-s2.0-S0260691716300764-main.pdf?_tid=190d2332-eb9f-4d41-8f07-27ce55881c59&acdnat=1531709549_d2df54c53d8ba579a8f759896740 4c48

References

- Children's Tumor Foundation. (2016). *NF1*. Retrieved from http://www.ctf.org/understanding-nf/nf1.
- Ferner, R.E., and Gutman, D.H. (2002). International consensus statement on malignant peripheral nerve sheath tumors in neurofibromatosis 1. *American Association for Cancer Research, 62*(5), 1573-1577. http://cancerres.aacrjournals.org/content/62/5/1573.article-info
- Neurofibromatosis Network (2018). What is NF. Retrieved from www.nfnetwork.org/understand-nf/what-is-nf/

APPENDIX E: PUBLIC GENOMIC AND GENETIC RESOURCES

Public genomic and genetic educational resources for health care professionals

Resource	Contact	Description
Centre for Education in Medical Genetics	http://www.bwhct.nhs.uk/ genetics-cemg-home.htm	Develops, provides, and evaluates genetics education opportunities and resources
Centre for Genetics Education	http://www.genetics.com.au/	Education and service resources for patients and professionals
Dolan DNA Learning Center	http://www.dnalc.org	Interactive, multimedia genetics education resources
Foundation for Genetic Education and Counseling	http://www.fgec.org	Educational resources on genetics and common diseases, especially psychiatric disorders (bipolar disorder and schizophrenia)
GenEd Project	http://www.medicine. man.ac.uk/GenEd/	Education and research links related to European aspects of genetic services
Genetics and Your Practice	http://www. marchofdimes.com/ gyponline/index.bm2	Online modules for healthcare professionals designed for exploration of a topic rather than sequential presentation of material . . . Many excellent fact sheets and sample clinical forms
Genetics in Clinical Practice: A Team Approach	http://iml.dartmouth.edu/ education/cme/Genetics/ or http://www.acmg.net/ resources/cd-rom-01/ intro.asp	Takes healthcare provider into a Virtual Genetics Clinic . . . Interactive virtual genetics clinic with case scenarios and case discussions . . . Target audience is primary care professionals
Genetics in Primary Care	http://genes-r-us. uthscsa.edu/resources/ genetics/primary_care.htm	Training program curriculum materials
Genetics in Psychology	http://www.apa.org/ science/genetics/ homepage.html	American Psychological Association's genetics site
Genetics Education Program for Nurses (GEPN) curriculum resources	http://www.cincinnati childrens.org/ed/ clinical/gpnf/default.htm	Sample genetics nursing course syllabi and other genetics educational opportunities and resources for nurses, as well as links to instructional resources used in GSI (Genetics Summer Institute) and WBGI (Web-based Genetic Institute)
Genetics: Educational	http://www.cincinnatichildrens.org/ed/clinicalgpnf/default.htm	Medical school course competencies, skills, knowledge,

Information		and behaviors which should be covered in genetics
Kansas Genetics Education Center	http://www.kumc.edu/ gec/	An ever-growing list of available resources, lesson plans, etc.
National Cancer Institute's CancerNet	http://www.cancer.gov/ cancerinfo/prevention genetics-causes	Authoritative information about cancer genetics
National Coalition for Health Professional Education in Genetics (NCHPEG)	http://www.kumc.edu/gec/	Core competencies in genetics and reviews of education programs . . . Descriptions of available instructional resources, courses, institutes . . . All have been submitted by developers and some have accompanying peer reviews
Physician's Database Query (PDQ®) Cancer Information Summaries	http://www.cancer.gov/ cancerinfo/pdq/genetics	PDQ® cancer information summaries in genetics
Practice-Based Genetics Curricula for Nurse Educators	http://www.fbr.org/ publications/pub_curic. html	Bound instructional modules with accompanying CD or PowerPoint presentations (sample chapter available online)
Six Weeks to Genomic Awareness	http://www.cdc.gov/ genomics/training/ sixwks.htm	Webcast of 12 segments of genomic topics for public health professionals

Retrieved from Health Professional Practice and Education, pg 63-65. American Nurses Association. (2009). *The essentials of genetic and genomic nursing: competencies, curricula guidelines, and outcome indicators, 2nd Ed.* [PDF file]. Retrieved from https://www.genome.gov/pages/careers/healthprofessionaleducation/geneticscompetency.pdf