Background

Educational Need and Purpose
Genetic considerations in primary care pediatrics are not limited to rare disorders or Mendelian traits. Genetics, genomics, and epigenetics play a profound role in health and disease and impact every child and family in a pediatric practice. The primary care pediatrician’s role is integral to the evaluation and management of patients with genetic conditions, potentially genetic-related conditions, and common disorders with a genetic basis, through the effective use of family health history, expert referrals, and genetic testing.

This case series will focus on a wide range of clinical topics that are encountered in pediatric primary care and that require the resident to “think genetically” in order to adequately diagnose and treat patients in the continuity clinic. Without genetic thinking, a pediatrician could be missing some critical pieces of the puzzle when caring for patients. Through these case exercises, we will demonstrate how easy it is to incorporate genetics into a primary care practice and how seamlessly the puzzle pieces then fall into place.

Incorporating genetics into primary care involves a shift of paradigm in medical thinking. It is not simply the “dermatologic approach” of matching physical characteristics to syndromes in a book or diagnosing disease through identification of a gene mutation. It involves thinking about the genetic makeup of each individual in every encounter to develop differential diagnoses for disease or for preventive counseling. In this scenario, child health providers will need to incorporate a new approach to primary care, with emphasis on certain skills, and also will need to be reeducated and updated on new knowledge in genetics. —Cheng TL. Primary care pediatrics: 2004 and beyond. Pediatrics. 2004;113(6):1802–1809

Objectives
Upon completion of the Think Genetics! Case Series, residents should be able to:

- Recognize the role of genetics, genomics, and epigenetics in health and disease.
- Identify the role of the primary care clinician in the identification and management of patients with genetic conditions using family health history, expert referrals, and genetic testing.
- Describe the importance of constructing a problem-based family history and three-generation pedigree when considering a diagnosis, prevention, or treatment plan in pediatric primary care.
- Apply a wide range of available resources, technologies, and tools that promote early identification, referral, and management of patients with genetic conditions.
ACGME Sub-competencies and Developmental Milestones

The 9-part case series addresses ACGME Sub-competencies and developmental milestones, and they are described within each case. Cumulatively, the following sub-competencies and developmental milestones are addressed:

**Patient Care**
- Gather essential and accurate information about the patient
- Provide transfer of care that ensures seamless transitions
- Make informed diagnostic and therapeutic decisions

**Medical Knowledge**
- Use of scientific studies related to patient’s health problems

**Practice-based Learning and Improvement**
- Identify strengths, deficiencies, and limits of one’s knowledge

**Professionalism**
- Professionalization
- Humanism
- Develop awareness of limitations to engage in help-seeking behaviors
- Recognize ambiguity in clinical medicine and respond appropriately

**Interpersonal and Communication Skills**
- Communicate effectively across a broad range of backgrounds

**Systems-based Care**
- Coordinate patient care within the health system
- Work in inter-professional teams to enhance safety and improve quality