Overall Competencies for the Pediatric Residency Training Program

GENETICS

Goals and Competencies

PC: Patient Care; MK: Medical Knowledge; PBLI: Practice-Based Learning & Improvement
IC: Interpersonal & Communication Skills; P: Professionalism; SBP: Systems-Based Practice

GOAL 1: **Normal Versus Abnormal (Genetics)**. Understand how to differentiate disorders associated with genetic predisposition or genetic disease from normal states or acquired disorders.

COMPETENCIES: (PC, MK, PBLI, IC, P, SBP)	**<25%**	**25-50%**	**50-75%**	**75-100%**
1. Identify key concepts related to gene structure, molecular genetic techniques, mutations, and common patterns of inheritance (autosomal dominant and recessive, x-linked recessive, multifactorial, and related to maternal age).		1	2	3
2. Describe common methods or genetic diagnosis, including genetic screening tests available, and identify resources for up-to-date information on this topic.		1	2	3
3. Explain the concept of malformation etiologies: chromosomal, multifactorial, teratogenic, due to intrauterine factors.		1	2	3
4. Perform a thorough physical examination with emphasis on identifying major and minor congenital anomalies which may be signs of underlying genetic syndromes.	1	2		3
5. Explain indications for testing in the primary care setting for genetic or metabolic disorders (e.g., for findings such as short stature, developmental delay, minor congenital anomalies, failure to thrive, seizures, family history suggestive of certain inherited conditions).		1	2	3
6. Collect an appropriate family history for genetic disorders and identify risks when present, and triage families appropriately for genetic counseling.		1	2	3
7. Discuss key concepts related to testing for carrier states and genes predisposing to cancer and adult onset disorders (e.g., Alzheimers).		1	2	3

GOAL 2: Conditions Requiring Urgent Referral. Understand how to recognize and respond to urgent and/or severe conditions related to genetics and inherited metabolic disorders.

COMPETENCIES: (PC, MK, PBLI, IC, P, SBP)	**<25%**	**25-50%**	**50-75%**	**75-100%**
Recognize, provide initial evaluation and counseling for, and identify resources for emergency consultation regarding:				
1. Infants presenting with symptoms that indicate the possibility of a severe inborn error of metabolism (e.g., metabolic acidosis, hyperammonemia, unexplained seizures).		1	2	3
2. Chromosomal abnormalities that require prompt diagnosis in the perinatal period (e.g., Trisomy 13, 18, 21).			1	2,3
3. Understand critical illness or death suggestive of metabolic disorder, requiring collection of tissue samples before or at time of death.		1	2	3
4. Developmental delay with signs or symptoms suggesting an underlying metabolic or genetic disorder.		1	2	3

GOAL 3: Conditions Requiring Complex Case Management: Understand the pediatrician's role in the long-term management of children with complex or unusual genetic or metabolic disorders.

COMPETENCIES: (PC, MK, PBLI, IC, P, SBP)	**<25%**	**25-50%**	**50-75%**	**75-100%**
1. For commonly encountered disorders (e.g., Trisomy 21, Turner's Syndrome, Fragile X, neurofibromatosis) list presenting signs and symptoms and identify principles of long-term management.		1	2	3
2. Provide primary care and participate as a team member in medical and educational planning for patients with genetic disorders.		1	2	3
3. Explain how to identify and use resources in one's community for diagnosis, genetic counseling, therapy, and psychosocial support of children with genetic defects and congenital anomalies.		1	2	3
4. Participate in the process of genetic counseling and then review the experience with an experienced genetic counselor.	1		2	3

GOAL 4: Prevention: Understand key concepts for prevention and early detection of genetic disorders.

COMPETENCIES: (PC, MK, PBLI, IC, P, SBP)	**<25%**	**25-50%**	**50-75%**	**75-100%**
1. List the disorders screened for in one's state neonatal screening program, provide initial counseling, and explain management for an infant with a positive neonatal screening test for each disorder.			1	2,3
2. Discuss public health strategies to reduce risk for genetic disorders and congenital anomalies (e.g., early identification and screening programs to detect disease and carrier states, prenatal care, genetic counseling).		1	2	3
3. Demonstrate commitment to collect appropriate screening histories, participate in neonatal screening programs, provide initial counseling, and utilize resources for genetic counseling.		1	2	3