Genetics Goals & Objectives	Objectives by Competency and Level of Training			Assessment Methods
	PL-1	PL-2	PL-3	
GOAL 1: Prevention, Counseling and Screening. Understand the role of the pediatrician in preventing genetic disease, and in counseling and screening individuals at risk for these diseases.	**Patient Care**: Provide routine genetic preventive counseling to all parents and patients that addresses: 1. Disorders identified in the neonatal screening program in one's state 2. Folic acid supplementation before and during pregnancy 3. Early and routine prenatal care and routine genetic screening for disease during pregnancy 4. Routine screening specific to certain ethnic groups 5. Avoidance of known teratogens during pregnancy (e.g. isotretinoin and alcohol), and reassurance about most substances that are not teratogenic	**Patient Care**: Provide regular genetic screening: 1. Screen for known familial genetic disease processes using the appropriate method. 2. Describe screening methods, including CVS, amniocentesis, maternal serum screening and high-definition ultrasound in women over the age of 35 or at risk for having a child with a specific genetic problem 3. Identify screening programs to detect disease and carrier states in family members	**Patient Care**: Provide prenatal and postnatal genetic preventive counseling to parents and patients with specific genetic conditions, addressing: 1. Genetic disorders with known or presumed inheritance patterns, based on a constructed pedigree 2. Expected course of known genetic disorders 3. Risk factors, including advanced maternal or paternal age and previous children with genetic conditions 4. Internet and other resources and support groups for known genetic disorders	Direct Observation Global Evaluation
GOAL 2: Genetic Conditions Followed by a General Pediatrician. Assist in diagnosis of genetic conditions and counseling of parents, under the supervision of a geneticist.	**Patient Care**: Discuss the presenting signs and symptoms for commonly encountered genetic disorders (e.g., Trisomy 21, Turner Syndrome, Fragile X, neurofibromatosis, spina bifida, Marfan syndrome, achondroplasia) and identify accepted guidelines for care	**Patient Care**: Provide primary care for and participate as a team member in medical and educational planning for a patient with a genetic disorder	**Patient Care**: 1. Perform a thorough physical examination on a child suspected of a specific genetic disorder, identifying major and minor congenital anomalies that could be signs of an underlying genetic syndrome 2. Develop a management plan for commonly encountered genetic disorders, identifying principles of long-term management, including use of disorder-specific growth charts and practice guidelines	Direct Observation Global Evaluation
	Medical Knowledge: 1. Describe general concepts that explain chromosome structure and spontaneous mutations, and molecular genetic techniques commonly used in diagnosis of genetic diseases 2. Describe common patterns of Mendelian vs. non-Mendelian inheritance (autosomal dominant and recessive, X-linked, multifactorial, and the effect of maternal and paternal age), and demonstrate the ability to construct a pedigree 3. Identify common diseases with known inheritance patterns and describe the mode of inheritance, including: cystic fibrosis, sickle cell anemia, Marfan syndrome, Huntington's Disease, neurofibromatosis, and familial cancer syndromes	**Medical Knowledge**: 1. Discuss unusual patterns of inheritance (mitochondrial defects, triplet repeat, imprinting) 2. Describe how well child care differs in a child with a genetic condition, e.g., use of specific growth charts for specific conditions and physical findings	**Medical Knowledge**: 1. Discuss unusual patterns of inheritance (mitochondrial defects, triplet repeat, imprinting) 2. Describe how well child care differs in a child with a genetic condition, e.g., use of specific growth charts for specific conditions and physical findings	Global Evaluation In-Training Exam
			PBLI Develop strategies to learn about future advances in the understanding of genetic disorders, in order to incorporate into one's practice improved screening, identification, counseling and management of such disorders	Global Evaluation EBM eval
		Systems-Based Practice Identify the indicators that would lead you to seek a genetics consult	**Systems-Based Practice** Discuss the ethical, legal, financial and social issues involved in genetic testing of children for genetic disorders that may present in adulthood, testing children for carrier status, and providing medical care for patients with known fatal disorders	360º eval Global Evaluation
GOAL 3: Conditions Requiring Urgent Referral (Genetics and Inborn Errors of Metabolism). Recognize and respond to urgent and/or severe conditions related to genetics and inherited metabolic disorders. 1. Infants presenting with symptoms that indicate the possibility of a severe inborn error of metabolism (e.g., metabolic acidosis, hyperammonemia, unexplained seizures, ketosis or hypoketosis, profound hypoglycemia) 2. Dysmorphic features found in chromosomal abnormalities that require prompt diagnosis in the perinatal period (e.g., Trisomy 13, 18, 21) 3. Unexplained critical illness or death suggestive of metabolic disorder, requiring collection of tissue samples before or at time of death 4. Developmental delay with signs or symptoms suggesting an underlying metabolic or genetic disorder 5. Physiologic changes or regression of milestones that suggest a possible metabolic etiology (e.g., urea cycle disorders, mitochondrial disorders, lysosomal storage diseases, abnormalities of organic/amino metabolism)	**Patient Care**: Explain the findings on clinical history and examination that suggest a known or potential genetic disorder or inborn error of metabolism	**Patient Care**: Identify, explain, provide initial management and support, and seek urgent referral when these conditions are suspected	**Patient Care**: Recognize immediate life-threatening complications associated with the diagnosis and treatment of inherited metabolic conditions. Refer for intensive care as indicated	Direct Observation Global Evaluation
	Medical Knowledge: Create a strategy to determine if the following presenting signs and symptoms are caused by genetic disease or an inborn error of metabolism and determine if the patient needs treatment or referral: •Developmental delay •Dysmorphic features •Poor feeding •Vomiting •Failure to thrive •Seizures •Short stature •Hearing loss •Cleft lip/palate •Respiratory disorders •Obesity •Skin lesions •Hypotonia •Unusual behavior	**Medical Knowledge**: Identify appropriate clinical and laboratory tests to help identify genetic diseases and inborn errors of metabolism. Explain the reason for the test to a family and interpret the results, with the assistance of a geneticist. The tests should include the following: •Chromosome analysis (both metaphase and prophase) and FISH testing for specific disorders •Plasma and urine amino acids, urine organic acids, ammonia level, venous pH, lactate, pyruvate, and blood acylcarnitine profile •Molecular testing for Fragile X •DNA mutational testing for selected disorders	**Medical Knowledge**: Identify appropriate clinical and laboratory tests to help identify genetic diseases and inborn errors of metabolism. Explain the reason for the test to a family and interpret the results, with the assistance of a geneticist. The tests should include newer and future technologies developed for detection of genetic disorders (e.g., microarray technology)	Global Evaluation In-Training Exam
	Interpersonal Skills and Communication Provide effective patient education, including reassurance, for condition(s) commonly seen on the inpatient service	**Interpersonal Skills and Communication** Engage patients and families in shared decision making	**Interpersonal Skills and Communication** 1. Develop effective strategies for teaching students, colleagues, other professionals and laypersons. 2. Role model effective communication skills in challenging situations	360º Feedback Global Evaluation
		Systems-Based Practice Identify written and internet resources to aid in diagnosing a genetic or inborn error of metabolism, using physical findings along with laboratory examination		360º Feedback Global Evaluation