Subspecialty Rotation: Genetics	
****All Goals and Objectives for this rotation are identical across all PL years****	
Primary Goals for this Rotation	**Competencies**
GOAL I: Prevention, Counseling and Screening (Genetics and Inborn Errors of Metabolism). Understand the role of the pediatrician in preventing genetic disease, and in counseling and screening individuals at risk for these diseases.	
1. Provide routine genetic preventive counseling to all parents and patients that addresses: a) Disorders identified in the neonatal screening program in one's state b) Folic acid supplementation before and during pregnancy c) Early and routine prenatal care and routine genetic screening for disease during pregnancy d) Routine screening specific to certain ethnic groups e) Avoidance of known teratogens during pregnancy (e.g. isotretinoin and alcohol), and reassurance about most substances that are not teratogenic	K, PC, IPC, P
2. Provide prenatal and postnatal genetic preventive counseling to parents and patients with specific genetic conditions, addressing: a) Genetic disorders with known or presumed inheritance patterns, based on a constructed pedigree b) Expected course of known genetic disorders c) Risk factors, including advanced maternal or paternal age and previous children with genetic conditions d) Internet and other resources and support groups for known genetic disorders	K, PC, IPC, P, SBP
3. Provide regular genetic screening: a) Screen for known familial genetic disease processes using the appropriate method. b) 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 c) Identify screening programs to detect disease and carrier states in family members	K, PC, SBP
GOAL II: Normal Vs. Abnormal (Genetics and Inborn Errors of Metabolism). Differentiate disorders in patients associated with genetic predisposition or genetic disease from normal states or acquired disorders.	
1. Describe general concepts that explain chromosome structure and spontaneous mutations, and molecular genetic techniques commonly used in diagnosis of genetic diseases.	K

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.	K
3. Discuss unusual patterns of inheritance (mitochondrial defects, triplet repeat, imprinting).	K
4. 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.	K
5. Identify common disorders with unusual inheritance patterns and describe the mode of inheritance, including: Fragile X, MERRF, and MELAS.	K
6. Explain the findings on clinical history and examination that suggest a known or potential genetic disorder or inborn error of metabolism.	K
7. 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.	K, PC
8. 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.	K, PC
9. 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: a) Chromosome analysis (both metaphase and prophase) and FISH testing for specific disorders b) Plasma and urine amino acids, urine organic acids, ammonia level, venous pH, lactate, pyruvate, and blood acylcarnitine profile c) Molecular testing for Fragile X d) DNA mutational testing for selected disorders e) Newer and future technologies developed for detection of genetic disorders (e.g., microarray technology)	K, PC, IPC, P
10. Identify written and internet resources to aid in diagnosing a genetic or inborn error of metabolism, using physical findings along with laboratory examination.	K, PC, PBLI
11. 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.	K, PC, P, SBP
12. 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.	K, PC, PBLI
13. Identify the indicators that would lead you to seek genetics consult.	K, PC
GOAL III: Undifferentiated Signs and Symptoms (Genetics and Inborn Errors of Metabolism). Evaluate, treat, and/or refer patients with the presenting signs and symptoms that suggest a genetic disease process.	
1. 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.	K, PC

a) Developmental delay b) Dysmorphic features c) Poor feeding d) Vomiting e) Failure to thrive f) Seizures g) Short stature h) Hearing loss i) Cleft lip/palate j) Respiratory disorders k) Obesity l) Skin lesions m) Hypotonia n) Unusual behavior	
GOAL IV: Genetic Conditions Followed by a General Pediatrician. Assist in diagnosis of genetic conditions and counseling of parents, under the supervision of a geneticist.	
1. 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.	K, PC
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.	K, PC, SBP
3. Provide primary care for and participate as a team member in medical and educational planning for a patient with a genetic disorder.	K, PC, IPC, P
4. Identify resources in your community for diagnosis, genetic counseling, therapy, and psychosocial support of children with genetic defects and congenital anomalies.	K, PC, SBP
GOAL V: 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. Identify, explain, provide initial management and support, and seek urgent referral for the following genetic and/or metabolic conditions: a) 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) b) Dysmorphic features found in chromosomal abnormalities that require prompt diagnosis in the perinatal period (e.g., Trisomy 13, 18, 21) c) Unexplained critical illness or death suggestive of metabolic disorder, requiring collection of tissue samples before or at time of death d) Developmental delay with signs or symptoms suggesting an underlying metabolic or genetic disorder e) Physiologic changes or regression of milestones that suggest a possible metabolic etiology (e.g., urea cycle disorders, mitochondrial disorders,	K, PC, IPC, SBP

lysosomal storage diseases, abnormalities of organic/amino metabolism)	
GOAL VI: Molecular Medicine. Recognize genetic factors in common diseases of childhood and adulthood	
1. Discuss current knowledge regarding the molecular basis of common childhood and adult conditions.	K
2. Identify the current and future uses of DNA testing in the office setting, including diagnosis of infectious diseases using DNA, pharmacogenetic testing for inborn errors of metabolic pathways prior to prescribing, DNA chips to identify genetic etiologies for complex disorders (e.g., congenital heart disease, seizure disorders, etc.).	K, PC, PBLI
3. Develop a strategy to incorporate concepts of molecular medicine into the everyday identification, treatment, counseling, and prevention of common disease processes.	K, PC, IPC
Procedures	
GOAL VII: Technical and therapeutic procedures. Describe the following procedures, including how they work and when they should be used; competently perform those commonly used by the pediatrician in practice.	K, PC
Wood's lamp examination of skin	

Core Competencies: **K** - Medical Knowledge
PC - Patient Care and Procedural Skills
IPC - Interpersonal and Communication Skills
P - Professionalism
PBLI - Practice-Based Learning and Improvement
SBP - Systems-Based Practice

Performance Expectations by Level of Training

	Beginning	**Developing**	**Accomplished**	**Competent**
	Description of identifiable performance characteristics reflecting a **beginning level** of performance.	Description of identifiable performance characteristics reflecting **development and movement toward mastery** of performance.	Description of identifiable performance characteristics reflecting **near mastery** of performance.	Description of identifiable performance characteristics reflecting the **highest level** of performance.
Medical Knowledge	PL1	PL1, PL2	PL2, PL3	PL3
Patient Care and Procedural Skills	PL1	PL1, PL2	PL2, PL3	PL3
Interpersonal and Communication Skills	PL1	PL1, PL2	PL2, PL3	PL3
Professionalism		PL1	PL2, PL3	PL3

Practice-Based Learning and Improvement	PL1	PL1, PL2	PL2, PL3	PL3
Systems-Based Practice	PL1	PL1, PL2	PL2, PL3	PL3

Milestones assessed on this rotation are:

Patient Care 1: History				
Level 1	**Level 2**	**Level 3**	**Level 4**	**Level 5**
Gathers information strictly following a template	Adapts template to filter and prioritize pertinent positives and negatives based on broad diagnostic categories or possible diagnoses	Filters, prioritizes, and synthesizes the history to develop a differential diagnosis in real-time for uncomplicated or typical presentations	Filters, prioritizes, and synthesizes the history to develop a differential diagnosis in real time for complicated or atypical presentations	Recognizes and probes subtle clues from patients and families; distinguishes nuances among diagnoses to efficiently drive further information gathering
Patient Care 4: Clinical Reasoning				
Level 1	**Level 2**	**Level 3**	**Level 4**	**Level 5**
Presents clinical facts (e.g., history, exam, tests, consultations) in the order they were elicited	Generates an unfocused differential diagnosis based on the clinical facts	Organizes clinical facts to compare and contrast diagnoses being considered, resulting in a prioritized differential diagnosis	Integrates clinical facts into a unifying diagnosis(es); reappraises in real time to avoid diagnostic	Role models and coaches the organization of clinical facts to develop a prioritized differential diagnosis, including life threatening diagnoses, atypical presentations, and complex clinical presentations
Interpersonal and Communication Skills 1: Patient and Family Centered Communication				
Level 1	**Level 2**	**Level 3**	**Level 4**	**Level 5**
Demonstrates respect and attempts to establish rapport	Establishes a therapeutic relationship in straightforward encounters	Establishes a culturally competent and therapeutic relationship in most encounters	Establishes a therapeutic relationship in straightforward and complex encounters, including those with ambiguity and/or conflict	Mentors others to develop positive therapeutic relationships
Attempts to adjust communication strategies based upon patient/family expectations	Adjusts communication strategies as needed to mitigate barriers and meet patient/family expectations	Communicates with sensitivity and compassion, elicits patient/family values, and acknowledges uncertainty and conflict	Uses shared decision making with patient/family to make a personalized care plan	Models and coaches others in patient- and family-centered communication