

APPENDIX A

2022 ITE Blueprint Specifications

	# Items
01 Basic Principles of Medical Genetics	65
A. Single gene inheritance	
A.01 Risk assessment	
A.02 General principles	
B. Population genetics	
C. Gene environment interactions	
D. Genomics	
E. Chromosome and gene structure and function	
F. Population screening	
G. Genetic counseling	
G.01 Ethical issues	
H. Chromosomal basis of inheritance	
I. Cytogenetics	
02 Cancer	22
A. Common familial and rare cancer syndromes	
03 Clinical Genetics (Pediatric and Adult)	31
A. Connective tissue disorders, skeletal dysplasias	
B. Hematologic disorders, cardiovascular disorders	
C. Neurogenetic, neuromuscular disorders	
D. Neurodegenerative disorders	
E. Ophthalmologic, hearing loss disorders	
F. Renal, pulmonary, gastrointestinal, immunologic disorders	
G. Pharmacogenomics	
H. Complex disorders (common adult disorders)	
I. Mechanisms of birth defects	
04 Reproductive Genetics	22
A. Reproductive genetics	
05 Multiple Malformations/Dysmorphology	30
A. Common dysmorphic syndromes	
B. Chromosomal disorders	
C. Intellectual disability, autism	
D. Congenital abnormalities	
E. Multifactorial disorders	
F. Long-term management	
06 Biochemical	30
A. Inborn errors of metabolism	
B. Newborn screening	
TOTAL	200