Lecture		Ch/Pg
1. Introduction & Genetic variation	• History • Review of basic principles in cell biology • Genetic variation (mutational & non mutational) • Detection of genetic variation	Ch1 & 2 REVIEW Ch3
2. Single Gene disorders I	• Autosomal dominant • Autosomal recessive • Penetrance and expressivity • Codominance • Dominant Negative • Examples (RB, Marfan, Cystic fibrosis) • Consanguinity	Ch4
3. Single Gene disorders II	• Lyonization • X-linked recessive (haemophilia) • X-linked dominant • Sex limited and influenced traits • Mitochondrial inheritance • Genomic imprinting (PWS & AS) • Anticipation & repeat expansion (Myotonic dystrophy, Fragile X)	Ch5
4. Clinical cytogenetics	• Karyotype, FISH, & CGH • Numerical chromosomal abnormality (trisomy 21,18,13; XO, XXY) • Structural chromosomal abnormality (22q11.2 deletion syndrome) • When do we order chromosomal analyses	Ch6

Lecture		Ch/Pg

5-6. Biochemical genetics	• Inborn errors of matabolism
	  • Carbohydrates & glycogen storage diseases
	  • Amino acids (PKU)
	  • Lipids & steroid hormones (MCADD)
	• Lysosomal storage diseases (Hurler, Gaucher, Tay-sachs…)
	• Urea cycle disorders
	• Energy production defects
	• Transport system abnormalities (Cystine & heavy metals)
		Ch7

7. Disease Gene identification	• Linkage analysis • Gene mapping • GWAS	Ch8
8. Multifactorial inheritance and common disease	• Principles • Nature vs Nurture • Common diseases	Ch12
9. Genetic testing & gene therapy		
10. Genetics & precision medicine		
11. Clinical genetics and genetic counseling		