For the genetic conditions listed below it is required to learn:
- key clinical features
- inheritance pattern
- gene (locus) and main diagnostic methods

Factor V Leiden Thrombophilia
Hemophilia type A
Hemochromatosis
Deletion 22q11.2 syndrome
Noonan syndrome
Williams syndrome
Neurofibromatosis type 1 (NF1)
Tuberous sclerosis
Joubert syndrome
Prader-Willi syndrome
Patau syndrome
Edwards syndrome
Down syndrome
Wolf-Hirschhorn syndrome
Charcot-Marie-Tooth neuropathy
Duchenne Muscular Dystrophy
Spinal Muscular Atrophy
Phenylketonuria
Deafness due to *GJB2* mutation (DFNB1)
Angelman syndrome
Fragile X syndrome
Huntington's disease
Rett syndrome
Cystic Fibrosis
Achondroplasia
Marfan syndrome
Klinefelter syndrome
Turner syndrome