Achondroplasia

Huntington's disease

Marfan syndrome

Hereditary spherocytosis

Polycystic kidney disease

Familial hypercholesterolemia

Sickle cell anaemia

Cystic fibrosis

Tay–Sachs disease

Phenylketonuria

Galactosemia

Haemophilia

Duchenne muscular dystrophy

hereditary multiple exostoses

tuberous sclerosis

Von Willebrand disease

Albinism

Tay–Sachs disease

Niemann–Pick disease

spinal muscular atrophy

Roberts syndrome

Rett syndrome

Down syndrome

Turner syndrome

Aicardi syndrome

Edward syndrome