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hydroxylase deficiency (AR); Porphyria variegata (AD); G6PD deficiency (X-linked R) (see Chapter 11); N-acetyl transferase deficiency (AR); Pseudocholinesterase deficiency (AR); Halothane sensitivity, malignant hyperthermia (genetically heterogeneous); Thiopurine methyltransferase deficiency (ACo-D); 5: Autosomal dominant inheritance, clinical examples; Overview; Disorders of the fibroblast growth factor receptors; Achondroplasia; Marfan syndrome (MFS); Familial hypercholesterolaemia (FH); Dentinogenesis imperfecta 1 (DGI) Otosclerosis 1 (OTSC1)Adult polycystic kidney disease (APKD, PKD); Multiple hereditary exostoses (EXT); 6: Autosomal recessive inheritance, principles; Overview; Rules for autosomal recessive inheritance; Example: albinism; Estimation of risk; Example: congenital deafness; 7: Consanguinity and major disabling autosomal recessive conditions; Overview; Management issues; Consanguineous matings; Incestuous matings; Brother-sister matings; Parent-child matings; Risk for offspring; First cousin marriages; Mental handicap; Oculocutaneous albinism; Recessive blindness; Retinitis pigmentosa (RP)
Severe congenital deafness Connexin 26 defects (CX26); Pendred syndrome (PDS); 8: Autosomal recessive inheritance, life-threatening conditions; Overview; Cystic fibrosis (CF); Tay-Sachs disease, GM2 gangliosidosis; Phenylketonuria (PKU); Spinal muscular atrophy (SMA); 9: Aspects of dominance; Overview; Codominance (Co-D), the ABO blood groups; Incomplete dominance, overdominance and heterosis; Incomplete penetrance; Delayed onset; Variable expressivity; Neurofibromatosis type 1 (NF1), Von Recklinghausen disease; 10: X-linked and Y-linked inheritance; Overview
Rules of X-linked recessive inheritance

Sommario/riassunto

Medical Genetics at a Glance covers the core scientific principles necessary for an understanding of medical genetics and its clinical applications, while also considering the social implications of genetic disorders. This third edition has been fully updated to include the latest developments in the field, covering the most common genetic anomalies, their diagnosis and management, in clear, concise and revision-friendly sections to complement any health science course. Medical Genetics at a Glance now has a completely revised structure, to make its content even