Genetic and Metabolic Disease in Pediatrics: Butterworths International Medical Reviews (Butterworth International Medical Reviews. Pediatrics)

June K. Lloyd

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Genetic and Metabolic Disease in Pediatrics is a compendium of papers that discusses the problems of inborn diseases in terms of homeostasis. One paper traces "backward" from the disease phenotype to discover and investigate the gene, as well as moves "forward" from mutation in DNA to discover phenotypes or proteins connected with the disease. Specific genes are assigned to particular places (loci) on chromosomes that can manifest the presence or type of disease. Another paper examines a classical disease-osteogenesis imperfecta-pointing out that the aberrant collagen of osteogenesis imperfecta reflects mutation at chromosomes 7 and 17. Another paper shows that in osteogenesis imperfecta, Mendelian phenotypes lead to genes and their products as being involved in critical aspects of protein traffic in human cells. Several papers examine the inborn errors of metabolism covering the lacticacidemias, urea synthesis, the hyperphenylalaninaemias, and the hyperlipidaemias. Other papers investigate the effects of metabolic dishomeostasis caused by variant maternal genotypes on fetal development, the "androgen pathway, its known Mendelian variants

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