This is a comprehensive and up-to-date survey of mental retardation caused by genes on the X chromosome.

Clinical and laboratory data on 130 syndromes are presented in a concise and consistent manner. Each syndrome is defined and information is provided on somatic features, growth and development, neurological signs, cognitive performance, imaging and other laboratory findings, and when possible, the nature and localization of the responsible gene. Craniofacial and other somatic findings are extensively illustrated. A differential matrix accompanies each syndrome description to assist the reader in identifying other X-linked syndromes with overlapping features. As background, the X chromosome and the emergence of the concept of X-linked mental retardation are discussed. X-chromosome maps show the location and mapping limits of the responsible genes. The authors have extensive experience in the clinical and laboratory delineation of X-linked mental retardation. They have described new syndromes, regionally mapped disease loci on the X chromosome, and in several cases isolated the genes responsible for X-linked syndromes.