Record Nr. UNINA9911019919503321 **Titolo** Variation in the human genome / / [editors, Derek Chadwick and Gail Cardew1 Chichester;; New York,: Wiley, 1996 Pubbl/distr/stampa **ISBN** 9786612347955 9781282347953 1282347950 9780470514887 0470514884 9780470514894 0470514892 Descrizione fisica 1 online resource (341 p.) Collana Ciba Foundation symposium:: 197 Altri autori (Persone) ChadwickDerek CardewGail Disciplina 573.2/1 Soggetti Human population genetics Human genome Human genetics - Variation Lingua di pubblicazione Inglese **Formato** Materiale a stampa Livello bibliografico Monografia Symposium on Variation in the Human Genome, held at the Ciba Note generali Foundation, London, 15 June 1995. Nota di contenuto VARIATION IN THE HUMAN GENOME: Contents: Participants: Introduction; Phylogeographic variability in traditional societies; Interpreting genetic variability: the effects of shared evolutionary history; Microsatellites: evolution and mutational processes; The phenylalanine hydroxylase locus: a marker for the history of phenylketonuria and human genetic diversity; Genetic and geographical

variability in cystic fibrosis: evolutionary considerations; Unusual inheritance patterns due to dynamic mutation in fragile X syndrome Control of b-thalassaemia by carrier screening, genetic counselling and prenatal diagnosis: the Sardinian experienceMultigenic control of skin tumour development in mice1; Population genetics of tumours; Genetic

factors that contribute to interindividual variations in plasma low

density lipoprotein-cholesterol levels1; Genetic architecture of common

mu Itifactorial diseases; World distribution of HLA alleles and implications for disease

Cladistic approaches to identifying determinants of variability in multifactorial phenotypes and tt e evolutionary significance of variation in the human genomeQuantitative phenotype analysis for localization and identification of disease-related genes in a complex genetic background; The genetics of common diseases: the implications of population variability; Final discussion; Summary; Index of contributors; Subject index

## Sommario/riassunto

The mapping of human genes is proceeding rapidly. Genes associated with specific inherited diseases are being identified, often providing insight into the molecular cause of the disease. At the moment, however, little consideration is being given to the variation present in different human populations. Variation in the Human Genome discusses methods of analysing population genetic data and how contemporary genetic heterogeneity arises during the evolution and migration of human populations. Specific disorders such as cystic fibrosis, beta-thalassaemia, fragile X, phenylketonuria and tumour dev