population cytogenetics STUDIES IN HUMANS

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Population Cytogenetics Studies In Humans

Harold Kalter

Population Cytogenetics Studies In Humans:

Population Cytogenetics Ernest B. Hook, Ian H. Porter, 1977 Population Cytogenetics ,1977 Atlas of Human Chromosome Heteromorphisms H.E. Wyandt, Vijay S. Tonk, 2013-03-09 Critical to the accurate diagnosis of human illness is the need to distinguish clinical features that fall within the normal range from those that do not That distinction is often challenging and not infrequently requires considerable experience at the bedside It is not surprising that accurate cytogenetic diagnosis is also often a challenge especially when chromosome study reveals morphologic findings that raise the question of normality Given the realization that modern human cytogenetics is just over five decades old it is noteworthy that thorough documentation of normal chromosome var tion has not yet been accomplished One key diagnostic consequence of the inability to distinguish a normal variation in chromosome structure from a pathologic change is a missed or inaccurate diagnosis Clinical cytogeneticists have not however been idle Rather progressive biotechnological advances coupled with virtual completion of the human genome project have yielded increasingly better microscopic resolution of chromosome structure Witness the progress from the early short condensed chromosomes to the later visualization of chromosomes through banding techniques hi resolution analysis in prophase and more recently to analysis by fluorescent in situ Individual Susceptibility to Genotoxic Agents in the Human Population Frederick J. De hybridization FISH Serres, Ronald W. Pero, William Sheridan, 2012-12-06 As a result of the industrial revolution man s technological achievements have been truly great increasing the quality of life to almost unimagined proportions but all this progress has not been accomplished without equally un imagined health risks Sufficiently diagnostic short term assay procedures have been developed in recent years for us to determine that there are mutagenic agents among thou sands of chemicals to which the human population is exposed today These chemicals were not significantly present prior to the indus trial revolution As of today there are no procedures available which have been adequately demonstrated to assess individual sus ceptibility to genotoxic exposures and as a result we have had to rely on extrapolating toxicological data from animal model systems The question is can we afford to allow such an increased environ mental selection pressure via mutagenic exposures to occur without expecting adverse long term effects on our health It is apparent from this line of reasoning that what is lacking and immediately needed are test procedures that can be applied to humans to assess genotoxic exposure as well as individual susceptibility to it There have already been two conferences which have focused at tention on this research area Guidelines for studies of human populations exposed to mutagenic and reproductive hazards A D Bloom ed March of Dimes Birth Defects Foundation White Plains New York 1981 and Indicators of genotoxic exposure in humans Banbury Report 13 B A Bridges B E Butterworth and I B Research Awards Index ,1989 Biomedical Index to PHS-supported Research ,1990

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formerly printed under the title Atlas of Human Chromosome Heteromorphism The Atlas has become a standard reference book in most cytogenetic laboratories and is cited as a significant reference in ISCN 2009 This revised version has updated and retained the most useful pictorial sections of the first edition including the comprehensive review of normal and not so normal variations of the human karyotype with summaries and extensive reference lists organized by chromosome number This updated edition features concise background information on chromosome methods and applications essential information on heteromorphism frequencies in normal and clinical populations as well as new listing and discussions of euchromatic subtelomeric and FISH variants The addition of two new sections make this an even more valuable reference than before A section on common and rare fragile sites includes a short historical discussion definitions and an extensive table of officially recognized sites that includes the HUGO name chromosomal location methods of induction genes and references to the most recent molecular characterization A new section on array CGH discusses the clinical challenge of interpreting copy number variations CNVs revealed by this newest technology gives examples of various levels of interpretation and lists the several most common websites used in this interpretation **Issues and Reviews in Teratology** Harold Kalter, 2012-12-06 Teratology is at once among the oldest and youngest of human preoccupations Coincident with man's first observations of the stars were his recordings of human and animal deformities But such aberrancies must have occurred even earlier for although it is one of those things like evolution that cannot be proven it is nevertheless indisputable that dysmorphogenesis must have occurred from the time complex forms of life first arose on our planet and that from the beginnings of human awareness our species was conscious of such happenings From the earliest recordings of this fascination with the form and meaning of abnormality a tortuous but continuous line extends to modern struggles to understand and control these manifestations And now after long occupying an honorable but peripheral place in the halls of philosophical and scientific pursuits teratology has quite suddenly come to take a prominent position at the hub of a complex crossroads of human concerns This shift in its fortune has taken several forms Fetal maldevelopment has become the concern of environmentalists activists of various persuasions indus trial organizations government agencies ethicists parents i e individuals and groups whose actions are impelled by apprehension Such motives are of course not without basis the trauma of thalidomide left a scar yet raw For still others clinicians academics experimentalists the upsurge in the interest in fetal mal development is at a different level and their pursuits are broad taking external agents as but one of the causes of defective development <u>Current Catalog</u> National Library of Medicine (U.S.), First multi year cumulation covers six years 1965 70

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