



# Human Chromosome Variation: Heteromorphism and Polymorphism

*Herman E. Wyandt, Vijay Tonk*

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Human Chromosome Variation: Heteromorphism and Polymorphism was 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.

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