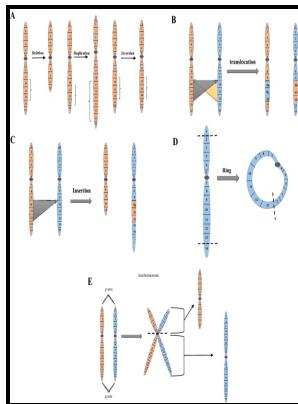


# Chromosomal variation in man - a catalog of chromosomal variants and anomalies

Wiley-Liss - Chromosome Anomaly



Description: -

- Chromosome Abnormalities -- indexes.
- Chromosome Aberrations -- indexes.
- Karyotypes -- Catalogs and collections.
- Human chromosomes -- Catalogs and collections.
- Human chromosome abnormalities -- Catalogs and collections.
- Chromosomal variation in man - a catalog of chromosomal variants and anomalies

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Notes: Includes bibliographical references (p. xxvii-xxviii) and indexes.

This edition was published in 1991



Filesize: 10.106 MB

Tags: #Y #to #X #chromosome #translocations

## Identification of balanced chromosomal rearrangements previously unknown among participants in the 1000 Genomes Project: implications for interpretation of structural variation in genomes and the future of clinical cytogenetics

The gene expression of each sample was compared to data reported for 13 EBV-B controls present in the Genotype-Tissue Expression GTEx project. Our previous pilot study showed the feasibility of detecting BCAs with low-pass or low-coverage paired-end WGS in a blinded fashion. The single-strand circle DNA ssCirc DNA library was rolling circle amplified to construct the DNA nanoball, which was substantially loaded into a patterned nanoarray.

### Chromosomal Variation in Man: A Catalog of Chromosomal Variants and Anomalies [Internet]

Contribute data: Genetic centres are invited to submit additional examples that may make it possible to establish whether the phenotypically normal abnormality carriers represent the benign end of a spectrum of phenotypic effect, or, whether there are indeed cytogenetic abnormalities that are consistently free of phenotypic consequences. A balanced chromosomal rearrangement or balanced chromosomal abnormality, BCA is a type of chromosomal structural variant SV involving chromosomal rearrangements e.

### Chromosomal Variation in Man: A Catalog of Chromosomal Variants and Anomalies [Internet]

Any mutation leading to an imbalance like monosomy, trisomy, or others can result in an abnormal phenotype. This results in an unbalanced chromosomal complement that will result in a zygote with partial trisomy of one chromosome and partial monosomy of the other when fertilized by a normal haploid gamete.

### Structural Changes in Chromosomes

Each reaction was performed in quadruplicate in 10 µl of reaction mixture simultaneously in cases and control DNA from YH EBV-B cell line on a StepOnePlus Real-Time PCR System Applied Biosystems with SYBR Premix Ex Taq II RNaseH Plus Takara Biotechnology, Dalian, China with the default setting of the reaction condition.

## **Chromosome Anomaly**

Because another sample type from this subject is not obtainable for further validation, this case NA18612 argues that peripheral blood is commonly considered a valuable sample type for disease studies for reasons beyond its simple availability. Am J Hum Genet 1983; 35:301—308. Nevertheless, five examples without direct transmission have been added to Group 1 including the dup 9 of Stumm et al 2002 , the del 10 of Davis et al 1999 , the dup 11 of Zarate et al 2007 , the dup 13 of Rivera et al 1981 and the dup 18 of Starke et al 2001.

### **Paracentric inversion in man**

One of these DNAs has a subtle translocation that is not readily identified by chromosome analysis because of the similarity of the banding patterns and size of exchanged segments, and another results in disruption of all transcripts of an OMIM gene. Calculation of insert size and read-pair quantities were based on nonchimeric and uniquely mapped read-pairs. This segregation pattern often is compatible with viability.

### **Structural Changes in Chromosomes**

Fluorescence in situ hybridization FISH was performed for NA18612 using standard procedures with bacterial artificial chromosome clones labeled by nick translation with SpectrumOrange, SpectrumRed or SpectrumGreen dUTP Abbott Molecular, Des Plaines, IL. The accessible chromatin landscape of the human genome.

### **Indexes & Databases**

Chromosome rearrangements can cause ill health in human population — infertility and mental retardation being the dominant effects.

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## Related Books

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