The human genome; you gain some, you lose some

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Kunst & Vliegwerk verzorgt een bijzondere vorm van dagbesteding voor kunstzinnig getalenteerde mensen met een verstandelijke handicap.

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Content

List of definitions

Acrocentric chromosomes:	Chromosomes lacking the short arm. The human acrocentric chromosomes are 13, 14, 15, 21, and 22.
Congenital malformation:	A physical defect present in the newborn.
Copy number:	The number of copies of a given chromosomal locus.
Copy number variation:	Alteration of a copy number of a certain DNA sequence in relation to the normal situation.
with phenotypic trait:	variation with clinical consequences.
without phenotypic trait:	variation without obvious clinical consequences (also called Polymorphic CNVs).
Deletion:	Loss of a DNA sequence.
Duplication:	An extra copy of a DNA sequence.
Duplicon:	Duplicon or segmental duplication has been defined as sequences of DNA greater than 1 Kb in size sharing a homology of at least 90 %.
False positive result:	An incorrect positive result of a test.
False negative result:	A result that appears negative but fails to reveal an alteration.
Gene:	Coding sequence.
Gene desert:	Region in the human genome that does not contain genes.
Genomic disorders:	The clinical condition that results from a dosage alteration of gene(s) located within a rearranged segment of the genome.
Mendelian inheritance:	Several inheritable traits or congenital conditions in humans are classical examples of Mendelian inheritance: Their presence is controlled by a single gene that can either be of the autosomal-dominant or -recessive type. People that inherited at least one dominant gene from either parent usually present with the dominant form of the trait. Only those that received the recessive gene from both parents present with the recessive phenotype (Wikipedia).

Mental retardation (MR) classification:	Mild MR (intelligent quotient (IQ) between 50 and 70), moderate MR (IQ between 35 and 50), severe MR (IQ between 20 and 35) and profound MR (IQ below 20).
Polymorphic CNVs:	CNVs (deletions as well as duplications) that are not related to a clinical phenotype (also called CNVs without phenotypic trait).
Phenotypic trait:	Any (abnormal) clinical feature, such as mental retardation, congenital malformations, dysmorphologies.
Translocations:	Exchange of genetic material between two different chromosomes.
Robertsonian translocations:	These translocations are produced by exchange in proximal short arms of the acrocentric chromosomes. Both centromeres are present, however, they function as one unit. This translocation is named after W.R.B. Robertson who described fusion of acrocentric chromosomes in insects.
Reciprocal translocations:	A translocation where part of one chromosome is exchanged with a part of a separate non-homologous chromosome.
Transposition:	Transfer of a segment of DNA to a new position on the same or another chromosome.
Uniparental disomy:	A euploid cell in which one of the chromosome pairs have been inherited exclusively from one parent. If two identical homologues are inherited this called isodisomy; if non-identical homologues are inherited the term heterodisomy is used. This occurs when non-disjunction during meiosis in one parent leads to formation of a disomic gamete. A trisomic zygote is formed and trisomic rescue with loss of the chromosome from the other parent occurs. UPD is of particular relevance in imprinted regions of the genome.

List of abbreviations

Bp	Base pair
BAC	Bacterial Artificial Chromosome
CGH	Comparitive Genome Hybridisation
CM	Congenital Malformation
CNVs	Copy Number Variation
COBRA	COmbined Binary RAtio
DD	Development Delay
DNA	Deoxyribonucleic acid
DOP-PCR	Degenerate Oligonucleotide Primed Polymerase Chain Reaction
FISH	Fluorescent in Situ Hybridisation
I.Q.	Intelligence Quotient
K	Kilo
Kb	Kilo base (one thousand base pairs)
LCR	Low Copy Repeat
MAPH	Multiplex Amplifiable Probe Hybridisation
Mb	Mega base (one million base pairs)
M-FISH	Multi-colour FISH
MLPA	Multiplex Ligation-dependent Probe Amplification
MR	Mental Retardation
NAHR	Non Allelic Homologous Recombination
Nt	Nucleotide
PAC	P1 derived Artificial Chromosome (PAC)
PCR	Polymerase Chain Reaction
PFGE	PulseField Gel Electrophoresis
RFLP	Restriction Fragment Length Polymorphism
SKY	Spectral Karyotyping
SNP	Single Nucleotide Polymorphism
UPD	Uniparental Disomy
VNTR	Variable Number of Tandem Repeats