

dominant gain  
of function

dominant loss  
of function

dominant neg-  
ative allele

autosomal rec-  
essive

compound het-  
erozygote

digenic inherit-  
ance

allelic heterog-  
eneity

allelic series

autosomal do-  
minant

<p>mutation in which the product interferes with normal production thus reducing function by &gt;50%</p>	<p>mutation which produces a 50% loss of function AA--&gt; Aa</p>	<p>mutation which produces a new function or increased function of product</p>
<p>phenotype requiring mutant genes at two loci</p>	<p>two alleles at same locus, both containing mutations. (a-1a2) aka heteroallelic</p>	<p>When heterozygote does not show recessive trait. Only seen in homozygous state. Not in every generation, equally between males and</p>
<p>When heterozygote shows dominant trait. Seen in every generation, equally between male and female (50%)</p>	<p>when different alleles at the same locus produce different phenotypes</p>	<p>different mutations at the same locus</p>

mosaicism

Penetrance

pleiotrophy

incomplete do-  
minance

locus heterog-  
eneity

modifying genes

genetic hetero-  
geneity

Heteroallelic

Homo allelic

<p>when one gene has consequences on many phenotypes</p>	<p>When a particular gene (mutation) does not always produce a phenotype (the probability that a mutation will show the mutant phenot-</p>	<p>Variable phenotypic expression (chimeric expression) on the cellular level (X inactivation)</p>
<p>when the product of a non allelic gene alters the expression of the phenotype associated with a particular gene (or mutation of</p>	<p>mutations in two genes at different loci which may produce a similar phenotype</p>	<p>when neither allele is dominant but rather phenotype is an intermediate</p>
<p>when both alleles are the same (homozygous)</p>	<p>compound heterozygous</p>	<p>similar phenotypes caused by different mutant genes</p>

gene deserts	homolog	ortholog
heterochromatin	conserved non-coding sequences (cNCS)	copy number variation (CNV)
pseudoautosomal	variable expression	euchromatin

<p>similar gene (w/ similar function) between species</p>	<p>genes which share a common ancestor</p>	<p>regions of genome devoid of genes- usually repetitive sequences and heterochromatin</p>
<p>sequence variation with large segments of genome that vary in number from one to several copies (ie genes in CNV region may exist in multiple copies)</p>	<p>areas of genome that are more conserved than would be expected by random mutation (total 5%, about 1.5% protein coding)</p>	<p>condensed regions of c/somes</p>
<p>uncondensed regions of c/somes can be transcriptionally active</p>	<p>degree to which a phenotype is expressed</p>	<p>regions on x and y c/some that are homologous</p>

LINES

SINES

Single nucleotide  
polymorphism  
(SNP)

haplotype block

linkage disequilibrium

Low copy  
repeats (LCR)

paralog

genomic disorders

haplotype

<p>Change in basepair sequence</p>	<p>Short (~300 bp) interspersed nuclear elements (created by insertions of transposable elements)</p>	<p>Long (~6 kb) interspersed nuclear elements (created by insertions of transposable elements)</p>
<p>region specific segmental duplication</p>	<p>the condition in which the haplotype frequencies in a population deviate from the values they would have if the genes at each locus were combined</p>	<p>the haplotype (group of genes) which is usually inherited as a unit</p>
<p>the genotype of a set of markers linked together on a segment of the same chromosome</p>	<p>problems/diseases which result from mutations in genome</p>	<p>homologous genes within an organism that belong to the same family but serve different functions</p>

linkage analysis	Short Tandem repeat polymorphism (STRP)	Susceptibility genes
genetic markers	genomic profiling	Relative risk (lambda)
Syntenic	Association Studies	complex traits

<p>makes an individual prone to a disease, but is not sufficient to cause a diseased phenotype</p>	<p>Short (200 bp) of tandem repeats of a simple 1-4 bp DNA sequence (aka microsatellites) can be used as markers</p>	<p>using markers that co-segregate with a phenotype, it is possible to find the gene associated with that phenotype</p>
<p>the recurrence risk for a relative of an affected person compared to the risk for a person from the general pop.</p>	<p>the detection of gene variants associated with a greater risk for a particular disease</p>	<p>sequences of the genome which can be inherited with particular genes. These serve to locate genes associated with a phenotype.</p>
<p>a phenotype that results from the interplay of multiple genes and the environment</p>	<p>compare the frequency of an allele in a population of affected individuals with the frequency of that allele in a population of unaffected individuals</p>	<p>genes on a contiguous piece of DNA-typically in a specific order</p>

homoplasmic

imprinting

isodisomy

anticipation

heterodisomy

heteroplasmic

Transmission  
disequilibrium test  
(TDT)

polygenic inhe-  
ritence

phenocopy

<p>A specific case of uniparental disomy when the pair results from duplication of the same chromosome</p>	<p>a case of stable transcriptional repression in which the silenced allele is determined by the parent of origin</p>	<p>when a cell contains only normal mtDNA</p>
<p>when a cell contains a mixture of mutant and normal mtDNA</p>	<p>A specific case of uniparental disomy when the pair results from inheritance of both homologs from one parent</p>	<p>apparent tendency of certain phenotypes to be increasingly severe in successive generations (pre-mutation)</p>
<p>when a phenotype is expressed for reasons unrelated to those with a specific genetic mutation (ie a person has disease X because of mutation in</p>	<p>variant alleles for multiple genes contribute to a particular phenotype</p>	<p>Comparing parents and affected offspring, the susceptibility allele (or marker) will be transmitted to offspring more often than the expected 1/2</p>

uniparental disomy	Mosaicism	
loss of imprinting (LOI)	parent of origin	epigenetics

	<p>The presence of at least two cell lines derived from a single zygote but differing in genotype and/or karyotype</p>	<p>when both members of a c/some pair are inherited from a single parent</p>
<p>"above" genetics-mitotically/meiotically heritable changes in gene expression not coded in DNA sequence</p>	<p>one parental allele may be preferentially expressed</p>	<p>When silenced gene is lost by malignant transformation making both genes active</p>