* Mendelian disease
* Non-coding variants (NCSs)
* Single nucleotide variants (SNVs)
* Chromosomal topological domains
* Next generation sequencing
* Structural variants (copy-number variants)
* Translocations
* ClinVar
* Splice sides
* Variant site
* Allel
* Acetylation
* Methylation
* Thimethylation
* SMOTE
* Cytogenetic Band
  + (One of the subregions of a chromosome visible microscopically after special staining. The cytogenetic bands are areas of the chromosome either rich in actively-transcribed DNA (euchromatin) or packaged DNA (heterochromatin).
* Chromosomal bands
* ReMM score
* Autosomen and Gonosomen
* Indel
* Dinucleotide bloc mutation
* Kb
* **Eutelic** organisms have a fixed number of somatic cells (any cell except sperm and egg cells) when they reach maturity
* Dna without nucleosome
* Ortholog genes
  + genes in different species that originated by vertical descent from a single gene of the last common ancestor.
* Evolutionary constraint
  + Cell lineage denotes
  + the developmental history of a tissue or organ from the fertilized embryo
* Conservation of gene order (known as syntheny)
* Transcription factors
  + proteins involved in the process of converting, or transcribing, DNA into RNA. Transcription factors include a wide number of proteins, excluding RNA polymerase, that initiate and regulate the transcription of genes.
* Chromatin accessibility
  + degree to which nuclear macromolecules are able to physically contact chromatinized DNA and is determined by the occupancy and topological organization of nucleosomes as well as other chromatin-binding factors that occlude access to DNA.
* Chromatin binding proteins, or DNA binding proteins
  + contain binding domains which are attracted to certain segments of DNA. These proteins are able to affect many aspects of DNA transcription and can be naturally occurring in cells or composed extracellular molecules.

Factor = Protein?

* Homeostatsis
  + state of steady internal, physical, and chemical conditions maintained by living systems
* Chromatin
* mit bestimmten Stoffen anfärbbarer Bestandteil des Zellkerns, der das Erbgut der Zelle enthält (‚Fäden‘ ohne Form vor der Zellteilung)
* Chromatid
* Teil der Chromosomen der Eukaryoten. Ein Chromatid besteht aus einem DNA-Doppelstrang und den zugehörigen Chromatin-Proteinen (ein / von X)
* A macromolecule
  + very large molecule, such as protein, commonly composed of the polymerization of smaller subunits called monomers. They are typically composed of thousands of atoms or more.
  + The four major biological molecules are nucleic acids, proteins, carbohydrates, and lipids. In this regard, a protein as a macromolecule is comprised of micromolecule monomers, amino acids linked together by peptide bonds.
* Peptides
  + short chains of between two and fifty amino acids, linked by peptide bonds. A polypeptide with more than fifty amino acids is a preotein.
* Homeotic genes
  + genes which regulate the development of anatomical structures in various organisms such as echinoderms, insects, mammals, and plants. Homeotic genes often encode transcription factor proteins, and these proteins affect development by regulating downstream gene networks involved in body patternin
* Transcription factor (TF).
  + A non- histone protein that directly binds to DNA.
* metazoan genomes
* Transposon (umgangssprachlich springendes Gen)
  + ist ein DNA-Abschnitt bestimmter Länge im Genom, der seine Position im Genom verändern kann (Transposition)
* Top down approach (genes)
* De novo sequence assemblers
  + are a type of program that assembles short nucleotide sequences into longer ones without the use of a reference genome. These are most commonly used in bioinformatic studies to assemble genomes or transcriptomes. Two common types of de novo assemblers are greegreedy algorithm assemblers and De Bruijn graph assemblers.
* De Bruijn graph
* Contigs
  + Continuous (or ‘contiguous’) sequences produced in a de novo assembly, free of any gaps.
* Scaffolds
  + Sets of ordered and oriented contigs with gaps.
* Euchromatin
  + Bereiche des aufgelockerten Chromatingerüsts im Karyoplasma einer Zelle dar. Im Gegensatz zum Heterochromatin liegt die Desoxyribonucleinsäure (DNA) hier in weniger dicht gepackter Form vor. Im Euchromatin befinden sich die meisten Gene und fast die gesamte Genaktivität. Zum Teil sind hier die Doppelstränge der DNA durch Enzyme zu parallelen Einzelsträngen aufgetrennt.
  + lightly packed form of chromatin (DNA, RNA, and protein) that is enriched in genes, and is often (but not always) under active transcription. Euchromatin comprises the most active portion of the genome within the cell nucleus. 92% of the human genome is euchromatic.
* String graph
* Read
  + inferred sequence of base pairs (or base pair probabilities) corresponding to all or part of a single DNA fragment. A typical sequencing experiment involves fragmentation of the genome into millions of molecules, which are size-selected and ligated to adapters. The set of fragments is referred to as a sequencing library, which is sequenced to produce a set of reads.
* Copy number variation (CNV)
  + phenomenon in which sections of the genome are repeated and the number of repeats in the genome varies between individuals
* Indel
  + a term for an insertion or deletion of bases in the genome of an organism.
* Missing heritability problem
* mitochondrial DNA
* Nucleosid = Base + Zucker
* Nukleotid = Nucleoside + Phosphatgruppe
* Nucleosom = viele Nukleotide um die acht Histone (146 Basenpaare)
* Haploinsufficient gene
  + is described as needing both alleles to be functional in order to express the wild type
* Paralogous genes (or paralogs)
  + are a particular class of homologous genes. They are the result of gene duplication and the gene copies resulting from the duplication are called paralogous of each other. After duplication, the paralogous genes can keep the same function, but they often diverge and develop different functions.
* Be in trans
* Compound heterozygosity
  + the condition of having two or more heterogeneous recessive alleles at a particular locus that can cause genetic disease in a heterozygous state; that is, an organism is a compound heterozygote when it has two recessive alleles for the same gene, but with those two alleles being different from each other (for example, both alleles might be mutated but at different locations)
* Heterozygous
  + having inherited different forms of a particular gene from each parent.
* Homozygous
  + having inherited the same alleles for a particular gene from both parents.
* Haplotype (haploid genotype)
  + A group of alleles in an organism that are inherited together from a single parent
  + a collection of specific alleles (that is, specific DNA sequences) in a cluster of tightly linked genes on a chromosome that are likely to be inherited together—that is, they are likely to be conserved as a sequence that survives the descent of many generations of reproduction.
  + a set of linked single-nucleotide polymorphism (SNP) alleles that tend to always occur together (i.e., that are associated statistically)
* Haploinsufficiency
* Homology directed repair
* Nonsynonymous (replacement mutations) substitutions
  + Biological changes, subject to natural selection
* Nearly neutral theory
* ABBA BABA Test
* Driver mutation
  + a mutation that gives a selective advantage to a clone in its microenvironment, through either increasing its survival or reproduction. Driver mutations tend to cause clonal expansions.
* Clone
  + a set of cells that all descend from a common ancestor cell. A clone is usually distinguished through inheritance of a distinctive genetic lesion (mutation) that occurred in the ancestor cell.
* Signatures (mutation)
* Clone = a set of cells that all descend from a common ancestor cell. A clone is usually distinguished through inheritance of a distinctive genetic lesion (mutation) that occurred in the ancestor cell.
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* ACGM - Advisory Committee on Genetic Modification
* Horizontal and vertical gene transfer
* Gene island

A single gene can produce multiple different RNAs (i.e., transcripts). The actual transcript observed will depend on the tissue, developmental time point, and environmental/hormonal/etc. factors. Typically, there's a single major transcript expressed in a given cell at a given time, but not always.

An engineered ‘dead’Cas9 (dCas9) variant with inactivating mutations in the endonuclease domains can be guided to bind specific DNA locations without cutting.

For some fungal organisms, such as Candida albicans, targeted gene inactivation is further hindered by their obligate diploid nature, and only partial homozygous KO libraries exist to date. WHY?

Occlude - verschließen

Susceptible - anfällig

Incongruous - unpassend

Excision

Endeavoring

Incipient – anfänglich

Indispensable – unverzichtbar

Repercussions Auswirkung

Rampant – wuchernd

Devoid – leer, ohne

Elucidation – Aufklärung

delineation – Beschreibung

miniscule – unbedeutend

scrutiny – Untersuchung

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