Medical Genetics

University Medicine · Vocabulary in context

Word	Example
medical genetics	**Medical genetics** studies how genes affect health.
karyotype	A **karyotype** is a picture of all 46 chromosomes. 46
meiosis /meɪˈəʊsɪs/	**Meiosis** makes sperm and egg cells.
nondisjunction /ˌnɒndɪsˈdʒʌŋkʃn/	In **nondisjunction**, chromosomes do not separate, so a cell gets one too many or too few.
germline mosaicism	In **germline mosaicism**, only some of a parent's eggs or sperm carry the change.
pedigree	A **pedigree** is a family tree drawn with squares for males and circles for females.
autosomal dominant	An **autosomal dominant** disease often appears in every generation.
autosomal recessive	An **autosomal recessive** disease can appear in children of healthy parents.
X-linked recessive	An **X-linked recessive** disease mostly affects boys.
X-linked dominant	An **X-linked dominant** disease needs only one changed X chromosome. X
carrier /ˈkærɪə/	A **carrier** has the allele but not the condition.
mitochondrial inheritance	In **mitochondrial inheritance**, genes pass only from the mother.

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genomic imprinting	In **genomic imprinting**, a gene is switched on or off depending on which parent it came from.
trinucleotide repeat expansion	A **trinucleotide repeat expansion** makes a three-letter DNA repeat grow longer. DNA
anticipation /ænˌtɪsɪpeɪʃn/	With **anticipation**, a disease starts earlier in each new generation.
synonymous	A **synonymous** change does not change the amino acid.
missense	A **missense** change swaps one amino acid for another.
nonsense	A **nonsense** change creates an early stop signal.
frameshift /ˈfreɪmʃɪft/	A **frameshift** changes every codon after the mistake.
splice	A **splice** site change can stop introns being cut out properly.
copy number variant	A **copy number variant** removes or doubles a large piece of DNA. DNA
variant of uncertain significance	A **variant of uncertain significance** is a DNA change whose effect we do not yet know. DNA
haploinsufficiency	In **haploinsufficiency**, one working copy of a gene is not enough.
Hardy–Weinberg equilibrium	**Hardy–Weinberg equilibrium** means allele frequencies stay the same over generations.

Word	Example
karyotyping	**Karyotyping** checks the number and shape of chromosomes.
microarray /ˈmaɪkrəʊəreɪ/	A **microarray** tests thousands of genes at once.
FISH /fɪʃ/	**FISH** uses glowing probes to find a piece of DNA on a chromosome. DNA
sequencing /ˈsiːkwənsɪŋ/	DNA **sequencing** reads the order of the bases. DNA
liability threshold model	The **liability threshold model** says disease appears when genes and environment add up past a limit.
Carter effect	The **Carter effect** says family risk is higher when the sex that is rarely affected has the disease.
pharmacogenomics	**Pharmacogenomics** uses a person's genes to choose the right drug.

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