



## Glossary

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#### **A2A receptors (also known as A2A and A2A receptor)**

signalling molecule found on brain cells and blocked by caffeine

#### **AAV**

a virus that can be used to deliver gene therapy drugs to cells. AAV stands for adeno-associated virus.

#### **acetyl**

a chemical tag that can be added to proteins or removed from them

#### **aggregate (also known as aggregates)**

Lumps of protein that form inside cells in Huntington's disease and some other degenerative diseases

#### **Allele**

one of the two copies of a gene

#### **alpha-1 (also known as alpha-1-antitrypsin)**

a protein which is faulty in a genetic disease called alpha-1-antitrypsin deficiency, which affects the liver and lungs

#### **amino acid (also known as amino acids)**

the building blocks that proteins are made from

#### **Amygdala**

A small brain area, in the temporal lobe, important for emotions and response to fear.

#### **amyloid**

The main protein that builds up in the brains of Alzheimer's disease patients

**anti-sense (also known as anti-sense strand)**

the half of the DNA double-helix that is mostly used as a backup, but sometimes produces message molecules

**Antioxidant**

a chemical that can 'mop up' harmful chemicals produced when cells release energy from food

**apoptosis**

A type of cell death where the cell uses specialized signals to kill itself

**ASOs (also known as Antisense oligonucleotides, Antisense oligonucleotide, and ASO)**

A type of gene silencing treatment in which specially designed DNA molecules are used to switch off a gene

**axon (also known as axons)**

long extensions of neurons, that act like electrical wires to carry signals in the nervous system.

**b****BAC**

an abbreviation for 'bacterial artificial chromosome'

**BACE1**

The gene for a protein called beta secretase 1, which thought to be involved in the development of Alzheimer's disease

**BACHD (also known as BAC-HD)**

a mouse model of Huntington's disease that develops symptoms slowly. BAC stands for bacterial artificial chromosome, referring to the way the HD gene was put into the mouse.

**BDNF**

brain-derived neurotrophic factor: a growth factor that may be able to protect neurons in HD

**biomarker**

a test of any kind - including blood tests, thinking tests and brain scans - that can measure or predict the progression of a disease like HD. Biomarkers may make clinical trials of new drugs quicker and more reliable.

**blood-brain barrier**

A natural barrier, made from reinforcements to blood vessels, that prevents many chemicals from getting into the brain from the bloodstream

**bone marrow**

The gooey stuff in the middle of bones, that manufactures blood cells. When eaten, gives dogs a healthy coat and vitality

## **C**

### **caffeine**

stimulant chemical found in tea, coffee and soft drinks like cola

### **CAG repeat**

The stretch of DNA at the beginning of the HD gene, which contains the sequence CAG repeated many times, and is abnormally long in people who will develop HD

### **Cas**

The bit of a gene-editing system that cuts the DNA. It's a type of protein called a nuclease. Cas is short for 'CRISPR-associated'.

### **caspase inhibitor (also known as caspase inhibition)**

A drug that reduces the activity of caspases, the enzymes that snip huntingtin protein up so it can enter the nucleus and cause problems

### **CED**

convection-enhanced delivery, a way of using pressure to spread drugs further in the brain

### **cervix**

the neck of the uterus (womb)

### **chaperone (also known as chaperones)**

chaperone proteins help other proteins to fold correctly, and can protect proteins against damage

### **chorea**

Involuntary, irregular 'fidgety' movements that are common in HD

### **chorionic villus sampling (also known as CVS)**

A medical procedure used to get a sample of DNA from a developing baby during pregnancy. A needle passed through the skin of the abdomen, or through the cervix, is used to collect tissue from the placenta.

**chromosomes**

Long strings of genes, tightly coiled into packages of DNA inside cells. Each cell's DNA is stored as 46 chromosomes. The HD gene is on chromosome 4. Each chromosome has two copies, one inherited from each parent.

**cilia**

Hair-like protrusions on the surface of cells

**circadian**

a circadian rhythm is something that repeats every day, like the body's sleep-wake cycle

**clinical trial**

Very carefully planned experiments designed to answer specific questions about how a drug affects human beings

**coenzyme-Q10**

a nutritional supplement that has some anti-oxidant properties

**cohort**

a group of participants in a clinical research study

**CRISPR (also known as CRISPR/CAS9)**

A system for editing DNA in precise ways

**cross-sectional study**

A study where each participant is looked at only once - unlike in a longitudinal study, where each participant is looked at several times over a time period

**CSF (also known as cerebrospinal fluid)**

A clear fluid produced by the brain, which surrounds and supports the brain and spinal cord.

**Cyclic-AMP**

a chemical found in neurons that responds to stimulation and is important for learning and memory

**Cyclic-GMP**

a chemical found in neurons that responds to stimulation and is important for learning and memory

**cytokine**

a signalling chemical of the immune system

**cytoplasm**

A part of a cell including everything inside the cell and outside the nucleus; where most of the cell's work happens

**d**

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**deep brain stimulation (also known as DBS)**

direct stimulation of the brain using electrical impulses through tiny wires.

**dendritic cell (also known as dendritic cells)**

A type of cell that comes from bone marrow and is part of the immune system

**differentiate (also known as differentiation)**

differentiation is the process of one cell type turning into another cell type.

**DNA fingerprinting**

a method of discovering what chromosome came from what parent or grandparent, without testing for the HD genetic mutation.

**dominant**

A genetic condition that only requires one copy of a mutation to occur

**dopamine**

A signaling chemical (neurotransmitter) involved in movement control, mood and motivation

**dystonia**

sustained involuntary muscle contractions, a bit like chorea but lasting longer

**e**

**efficacy**

A measure of whether a treatment works or not

**embryo (also known as embryos)**

the earliest stage during the development of a baby, when it consists of just a few cells

**Endoplasmic Reticulum**

part of the cell that, among other things, stores excess calcium

**enteric nervous system**

The collective name for the neurons found in the stomach and intestines.

**ependymal cells**

Cells that line the cavities of the brain

**excitotoxic (also known as excitotoxicity)**

the death of neurons due to overstimulation

**exclusion testing**

an optional add-on to prenatal testing, where DNA from parents and grandparents is compared with the DNA of the embryo or fetus. Exclusion testing means that the at-risk parent doesn't have to have an HD genetic test to have HD-free children.

**Exons (also known as exon and Exon)**

The small fraction of our DNA that is directly used to instruct cells how to make proteins

**exosome (also known as exosomes)**

Tiny bubble-like particles produced by cells that can transfer chemicals to other cells

**expression profiling (also known as gene expression profiling)**

a technique that allows measurement of the activation levels of many thousands of genes at once

**f****fetus**

a developing baby in the womb

**Food and Drug Administration**

The government regulatory authority in the US responsible for approving new drugs

**frontotemporal dementia**

a degenerative brain disease that can cause problems with speech and behavior

**g****GABA**

A chemical the brain uses to signal 'slow down' from one brain cell to another

**Ganglioside**

a type of fat with important roles in the brain, decreased in HD patient brains

**GDNF**

glial cell-derived neurotrophic factor: a growth factor that protects neurons in Parkinson's Disease, and maybe HD

**gene silencing**

An approach to treating HD that uses targeted molecules to tell cells not to produce the harmful huntingtin protein

**genetic engineering**

A technique used by scientists to alter the genes (DNA) of an animal or other organism, so that its cells produce different proteins and behave differently

**genome**

the name given to all the genes that contain the complete instructions for making a person or other organism

**Genome Editing**

The use of zinc-finger nucleases to make changes in DNA. 'Genome' is a word for all the DNA we each have.

## **glossary**

These popups will explain any technical terms we have to use.

### **glutamate**

a signalling chemical in the brain, or 'neurotransmitter'

### **glutamine**

the amino acid building block that is repeated too many times at the beginning of the mutant huntingtin protein

### **growth factor**

chemicals produced by the brain that help neurons to survive

## **h**

### **HDAC (also known as histone de-acetylase and HDACs)**

histone de-acetylases (HDACs) are machines that remove acetyl tags from histones, causing them to release the DNA they're attached to

### **heat shock response**

a defensive strategy that cells can activate when in danger or under stress. The heat shock response includes the production of more chaperone proteins, to help protect other proteins.

### **hippocampus**

the seahorse-shaped part of the brain that's crucial for memory

### **histone (also known as histones)**

special proteins that our DNA wraps around to stabilize and protect it

### **hormone (also known as hormones)**

Chemical messengers, produced by glands and released into the blood, that alter how other parts of the body behave

### **HTT**

one abbreviation for the gene that causes Huntington's disease. The same gene is also called HD and IT-15

### **HTTAS**

the anti-sense (backwards) version of the HTT gene

### **huntingtin protein**

The protein produced by the HD gene.

### **Hydrophilic**

## **hydrophilic**

'water loving' - chemicals that mix well with water are hydrophilic

## **Hydrophobic**

'water hating' - chemicals that do not mix well with water are hydrophobic

## **hypothalamus**

A tiny brain region with important roles in controlling the body's hormones and metabolism

## **i**

### **In vitro fertilization (also known as IVF and in-vitro fertilization)**

A medical procedure where eggs and sperm are combined in the laboratory, then embryos are implanted in the mother's womb.

### **induced pluripotent stem cells (also known as IPS cells)**

Stem cells that have been grown from adult cells.

## **inflammation**

Activation of the immune system, thought to be involved in the HD disease process

## **insulin**

A hormone that regulates the body's use of sugar and fats, and many other aspects of metabolism

## **intermediate allele**

A copy of the huntingtin gene with a CAG-repeat length that never leads to disease for the carrier, but may place the carriers children at some risk of HD.

## **intermediate alleles**

HD genes with CAG lengths between 27-35, which do not result in HD symptoms, but are longer than normal. Intermediate alleles are thought to be at risk of expanding in the offspring of people who carry them.

## **interventional**

A study in which an experimental drug or treatment is given to volunteers

## **intracranial volume**

An MRI measurement that reflects the largest size a brain ever achieves

## **j**

### **JM6**

an experimental drug that is converted by the body into Ro-61, which inhibits the enzyme KMO

### **juvenile HD (also known as JHD)**



Huntington's disease where symptoms begin before the age of 20.

## **k**

### **kinase**

A protein whose job it is to add a specific kind of chemical tag to other proteins. Kind of like a stapler.

### **KMO**

kynurenine mono-oxygenase, an enzyme that controls the balance of harmful and protective chemicals resulting from the breakdown of proteins

### **knock-in**

an organism that has had one of its genes altered, for example by adding a long CAG repeat into the huntingtin gene.

### **knockout**

a form of genetic engineering in animals are bred to lack a chosen gene

### **kynurenic acid (also known as Kyna)**

kynurenic acid, a chemical produced naturally in the brain that can protect neurons from harmful over-stimulation

### **kynurenine**

a chemical similar to kynurenic acid (Kyna)

## **l**

### **leptin**

A hormone that regulates appetite

### **longitudinal study**

A study where each participant is looked at several times over a time period - unlike a cross-sectional study, where each participant is looked at only once

## **m**

### **magnetic resonance (also known as MRI)**

A technique using powerful magnetic fields to produce detailed images of the brain in living humans and animals

### **manifest**

after HD diagnosis, or when symptoms are already showing

### **medium spiny neuronmedium spiny neurons**

brain cells in the striatum that are most damaged early in HD

### **Melatonin**

**melatonin**

a hormone produced by the pineal gland, important for regulating sleep

**Mesenchmal**

mesenchymal stem cells are cells with multiple functions that can be grown from adult bone marrow.

**messenger RNA (also known as mRNA)**

A message molecule, based on DNA, used by cells as the final set of instructions for making a protein.

**meta-analysis**

Combining the results of several different studies and analyzing them together, to increase their ability to answer a particular question.

**metabolism**

The process of cells taking in nutrients and turning them into energy and building blocks to build and repair cells.

**Metabolite (also known as metabolites)**

a chemical produced by cells as they break down fuels for energy

**Metabolomics**

the simultaneous measurement of many metabolites in a sample

**methionine**

one of the amino acid 'building blocks' that proteins are made from

**microdialysis**

a technique that enables researchers to measure the levels of chemicals in the living brain

**microglia**

the brain's immune cells

**mitochondria**

tiny machines inside our cells that process fuel into energy, enabling cells to function

**motor neuron disease**

A progressive neurological disease in which motor (movement) neurons die. Also known as ALS or Lou Gehrig's disease.

**multiple sclerosis (also known as MS)**

a disease of the brain and spinal cord, in which episodes of inflammation cause damage. Unlike Huntington's disease, MS isn't genetically inherited.

**n****n-terminal fragment**

A piece of a protein, like huntingtin, taken from the beginning of the protein.

**neurodegenerative (also known as neurodegeneration)**

A disease caused by progressive malfunctioning and death of brain cells (neurons)

**neuron (also known as neurons)**

Brain cells that store and transmit information

**neuroprotection (also known as neuroprotective)**

something that protects brain cells against damage

**neutron (also known as neutrons)**

Small sub-atomic particles that can be produced by some nuclear reactions and can help to study proteins in detail

**Non-disclosure**

an optional add-on to PGD, where an HD genetic test is performed on an at-risk parent but the result is kept secret. Non-disclosure PGD enables HD-free embryos to be implanted without the at-risk parent having to learn their HD status.

**Non-invasive prenatal diagnosis (also known as Noninvasive prenatal diagnosis and NIPD)**

A technique for preventing HD from being passed to children. A blood sample from the mother is taken, and DNA from the fetus is found in the blood and genetically tested for the risk of HD.

**nucleotide bases**

A single 'letter' of the genetic code, abbreviated A, T, G or C

**nucleus**

A part of the cell containing genes (DNA)

**O**

**observational**

A study in which measurements are made in human volunteers but no experimental drug or treatment is given

**open label**

A trial in which the patient and doctor know what drug is being used. Open label trials are susceptible to bias through placebo effects.

**p****PACSIN**

a normal protein that might be involved in helping huntingtin to function

**Parkinson's Disease**

A neurodegenerative disease that, like HD, involves motor coordination problems

**PDE10**

a brain protein that may be a good drug target and biomarker in Huntington's disease. PDE10 is found almost exclusively in parts of the brain where brain cells die in HD.

**peptides**

Small pieces of protein that signal in the body from one cell to another

**perseveration**

The inability to to change thoughts or actions to match changed plans

**phase III**

The phase in the development of a new treatment where clinical trials are conducted using many patients, to determine whether the treatment is effective

**phenylbutyrate**

a 'non-selective' HDAC inhibitor that affects all HDACs without targeting any particular HDAC enzyme

**phosphatase**

A protein whose job it is to remove a specific kind of chemical tag from other proteins. Kind of like a staple remover.

**Phosphodiesterase**

a protein that breaks down cyclic-AMP and cyclic-GMP

**pineal (also known as pineal gland)**

a gland in the brain that produces the hormone melatonin

**placebo**

A placebo is a dummy medicine containing no active ingredients. The placebo effect is a psychological effect that causes people to feel better even if they're taking a pill that doesn't work.

**placenta**

the 'afterbirth', which supplies the fetus with oxygen and nutrients via the umbilical cord.

The placenta's DNA is the same as the fetus's.

### **pluripotency**

the ability of some cells to turn into different types of cell

### **polymerase chain reaction (also known as PCR)**

PCR or the polymerase chain reaction is a scientific technique used to detect and amplify specific bits of DNA. It's used in the process of genetic testing for Huntington's disease.

### **PolyQ (also known as polyglutamine)**

A description of HD and other diseases that are caused by abnormal expansion of stretches of DNA containing the sequence CAG repeated many times. Too many CAGs in a gene results in proteins with too many 'glutamine' building blocks, and glutamine is represented by the symbol Q.

### **pooping**

Come on, you know what this one means

### **post-translational modification**

the addition of small chemical tags to a protein after it has been synthesized. These tags often change the location or function of the tagged protein.

### **Pre-implantation genetic diagnosis (also known as PGD)**

A technique for preventing HD from being passed to children. Eggs and sperm are combined in a laboratory, and the embryos are tested genetically for the mutation. Only embryos without it are implanted into the mother's womb.

### **Preimplantation genetic testing or PGT (also known as PGT and Preimplantation genetic testing)**

A technique for preventing HD from being passed to children. Eggs and sperm are combined in a laboratory, and the embryos are tested genetically for the mutation. Only embryos without it are implanted into the womb.

### **prenatal testing**

A technique for preventing HD from being passed to children. A DNA sample is taken during pregnancy and tested genetically. If the HD mutation is found, the pregnancy is terminated.

### **prevalence**

A figure estimating how many people there are in a particular population who have a certain medical condition.

### **primary endpoint**

The main question asked in a clinical trial

### **primate**

a group of mammal species including monkeys, apes and humans

**prion (also known as prions)**

special proteins that can become harmful, and cause disease called prion disease. Like falling dominoes, prion proteins can 'infect' other proteins, making them become harmful.

**prodromal**

prior to onset or diagnosis of movement symptoms

**promoter**

a special part of a gene devoted to turning the gene on and off

**putamen**

part of the striatum, a deep brain region important for movement control, that's affected early in the course of HD.

**q****quinolinic acid (also known as Quin)**

quinolinic acid, a chemical produced naturally in the brain that can harm neurons by overstimulating them

**r****R6/2**

a mouse model of Huntington's disease. R6/2 mice have been genetically altered with an abnormal gene that makes them produce a harmful fragment of the mutant huntingtin protein

**rabies**

A virus that infects the brain

**Receptor**

a molecule on the surface of a cell that signalling chemicals attach to

**recombinant**

artificial DNA that has been stuck together from small pieces from different sources.

**reduced penetrance allele**

A copy of the huntingtin gene with a CAG-repeat length long enough to cause symptoms in some, but not all, people who carry it.

**Rhes**

a protein found in the brain regions damaged early in Huntington's disease.

**ribosome**

A molecular machine that makes proteins using the genetic instructions in RNA message molecules

## **RNA**

the chemical, similar to DNA, that makes up the 'message' molecules that cells use as working copies of genes, when manufacturing proteins.

## **RNA interference (also known as RNAi)**

A type of gene silencing treatment in which specially designed RNA molecules are used to switch off a gene

## **Ro 61-8048 (also known as Ro-61)**

an experimental drug that inhibits the enzyme KMO

## **Ryanodine Receptor**

a pore that lets calcium enter cells

## **S**

### **SAHA**

an HDAC-inhibitor drug. Its full name is Suberoylanilide hydroxamic acid.

### **SBMA**

Spinobulbar muscular atrophy, another neurodegenerative disease caused by increased CAG size

### **SCA**

Spinocerebellar ataxia, another neurodegenerative disease caused by increased CAG size

### **secondary endpoints**

Additional questions asked in a clinical trial that help scientists look at treated patients as broadly as they can to determine the effects of a drug

### **sense strand**

the half of the DNA double-helix that contains instructions for most proteins. The 'business' strand.

### **significance testing**

A method used by statisticians to try to decide whether the result of a study or trial is genuine or likely to have happened by chance

**single nucleotide polymorphisms (also known as SNP)**

a single-letter spelling difference in a gene. SNPs, pronounced 'snips', are common and most don't change the function of the gene.

**siRNA**

A way of silencing genes using specially designed molecules of RNA – like DNA but made of only a single strand – that target the message molecules in cells and tell them not to make a certain protein

**Sodium**

A chemical element found abundantly on planet earth, in rocks, plants and animals (including humans). The main constituent of salt, AKA sodium chloride.

**soma**

the main cell body of a neuron, which contains the cell nucleus where genes (DNA) are located

**somatic**

relating to the body

**spinocerebellar ataxia**

A family of diseases which result in characteristic movement disorders. Many types of spinocerebellar ataxia are caused by the same type of mutation as HD – a CAG expansion.

**splicing**

the cutting up of RNA messages, to remove non-coding regions and join together coding regions.

**statistically significant**

Unlikely to have arisen by chance, according to a statistical test

**stem cells**

Cells that can divide into cells of different types

**suprachiasmatic nucleus (also known as SCN)**

the part of the brain that controls daily or 'circadian' rhythms

**Synapse**

the site of connection between two neurons in the brain

**Synaptic transmission**

The passage of messages between neurons at a synapse

**synaptophysin**

a protein essential for the formation of synapses, the connections between neurons.

**t**



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## **therapeutics**

treatments

## **Total Functional Capacity (also known as TFC)**

A standardized rating scale for function in HD, used to assess capacity to work, handle finances, perform domestic chores and self-care tasks

## **TR-FRET**

“Time Resolved Fluorescence Resonance Energy Transfer” - a sophisticated technique for measuring interactions between antibodies.

## **transcranial magnetic stimulation**

applying magnetic fields to the brain to study its functioning

## **transcription**

the first step in making a protein from the recipe stored in a gene. Transcription means making a working copy of the gene from RNA, a chemical messenger similar to DNA.

## **transcription factor**

a gene-control protein. In response to signals from inside and outside cells, transcription factors attach to the DNA and cause specific genes to be more or less activated, producing more or less of the corresponding protein.

## **transcriptional dysregulation**

disruption of the mechanisms that control the activation levels of different genes, leading to genes being wrongly activated or inactivated

## **transcriptional regulation**

the mechanisms that control the activation levels of different genes

## **transgenic**

an organism that has had an extra ‘foreign’ gene or genes inserted into its DNA.

## **trinucleotide repeat**

A stutter in the genetic code that results in the repetition of a three ‘letter’ sequence

# **U**

## **UHDRS**

A standardized neurological examination that aims to provide a uniform assessment of the clinical features of HD

## **uterus**

womb

# **V**

## **Vascular system**

the vessels and/or tissue that carry fluids like blood throughout the body

## **ventricle (also known as ventricles)**

Normal fluid-filled spaces within the brain.

## **vesicle**

a tiny 'bubble' produced by a cell that can deliver chemicals to other cells

# **W**

## **Whole Exome Sequencing (also known as whole exome sequencing and Whole exome sequencing)**

Decoding exons, the ~1.5% of a persons DNA which contains instructions for directly building proteins

## **Whole genome sequencing (also known as whole genome sequencing and Whole Genome Sequencing)**

Decoding every one of the approximately 3 billion 'letters' of an individuals DNA

## **wild-type**

the opposite of 'mutant'. Wild-type huntingtin, for example, is the 'normal', 'healthy' protein.

# **y**

## **YAC**

an abbreviation for 'yeast artificial chromosome'

## **YAC128**

one of several mouse models of HD. YAC stands for "yeast artificial chromosome".

# **Z**

## **Zinc-finger Nucleases**

Molecular machines that attach to a specific DNA sequence and then cut the DNA strand

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