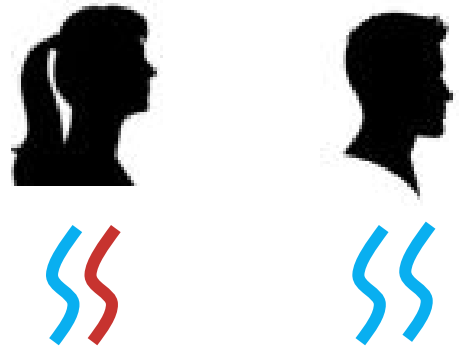
Huntington's Disease Education Day

HDSA Centre of Excellence
University of Washington, Tacoma

HDBUZZ

SKYHAWK THERAPEUTICS

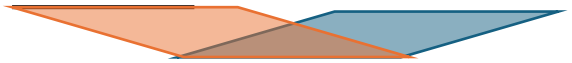
Alnylam PHARMACEUTICALS



uniQure

PTC THERAPEUTICS

NOVARTIS



Spark

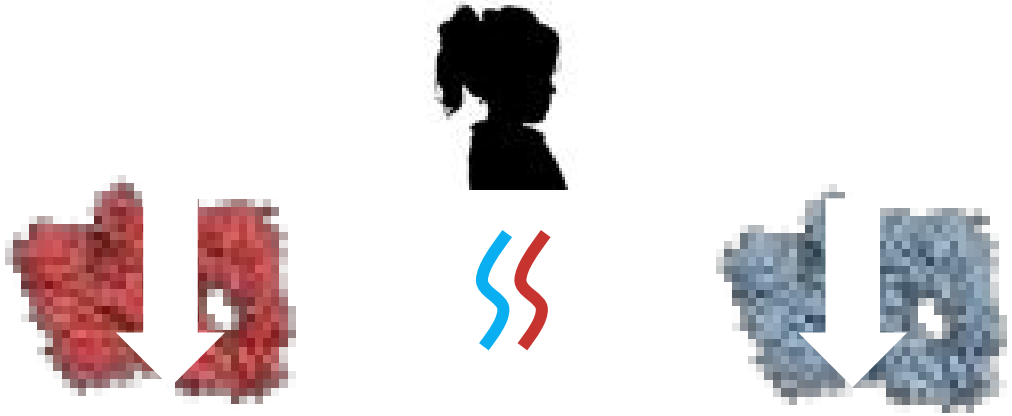
Roche

Genentech

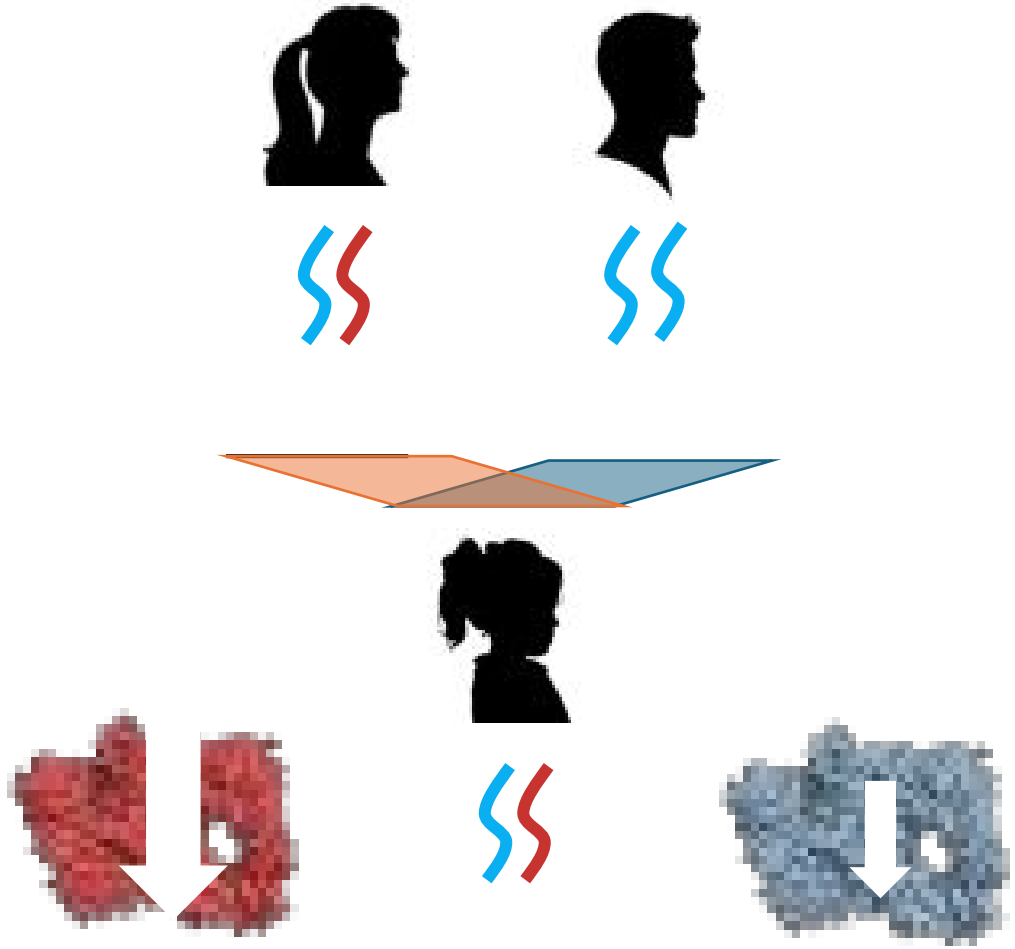
Roche

Genentech

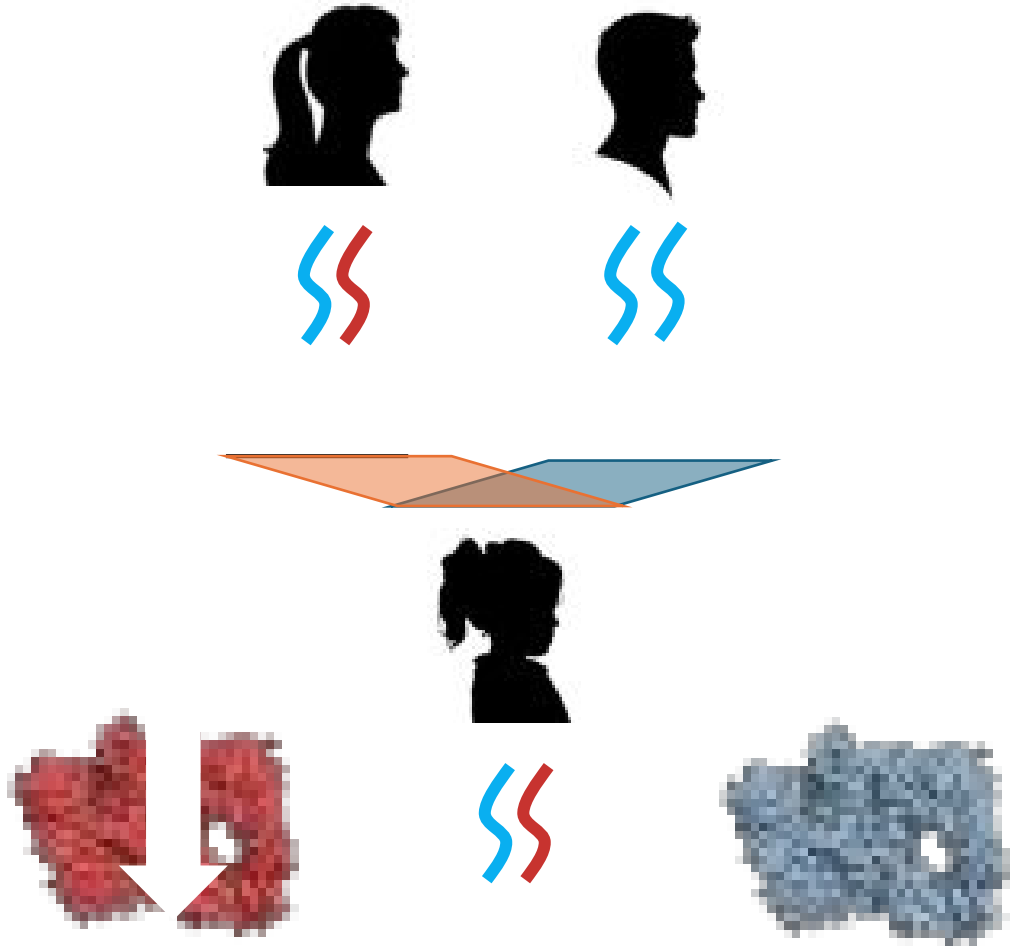
A Member of the Roche Group



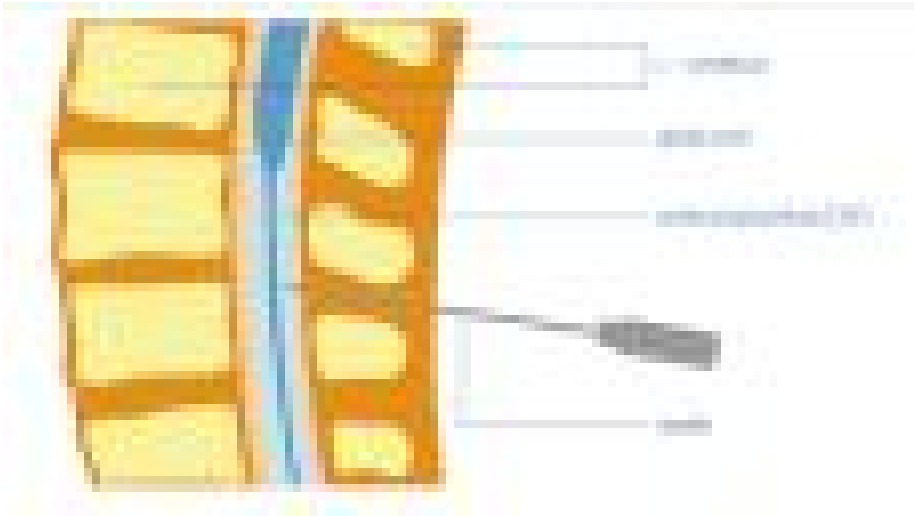
VICO Therapeutics

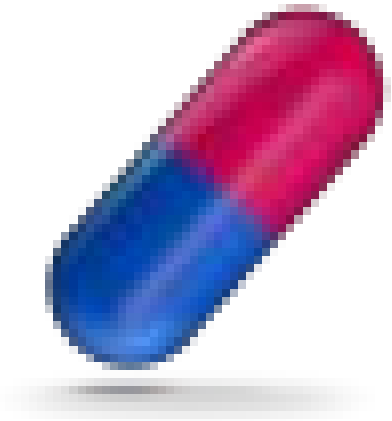


WAVE®
LIFE SCIENCES



HDBUZZ





PTC THERAPEUTICS

NOVARTIS

SKYHAWK THERAPEUTICS

Roche

Genentech
A Member of the Roche Group

WAVE LIFE SCIENCES

Alnylam PHARMACEUTICALS

VICO Therapeutics

uniQure

Spark THERAPEUTICS

Promising results in a small group of people

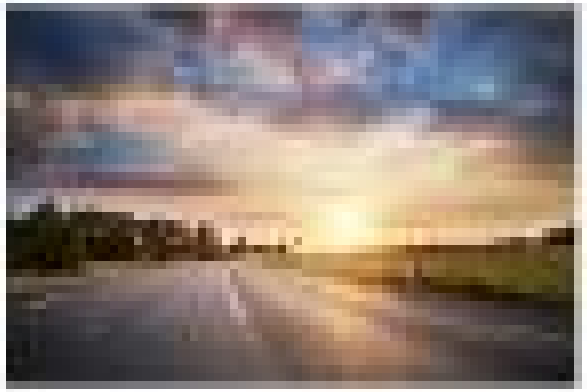
- 12 people on high dose of AMT-130 at 3-year point
- Primary endpoint met - 75% slowing of progression (cUHDRS)
- Secondary endpoint met - 60% slowing of TFC
- Compared to external control (data from Enroll-HD)

uniQure

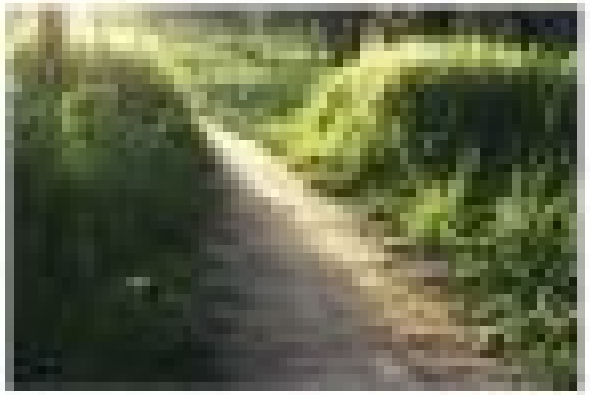
The path forward for AMT-130 has been bumpy

HDBUZZ

The road ahead for uniQure: FDA says more data needed for AMT-130 gene therapy

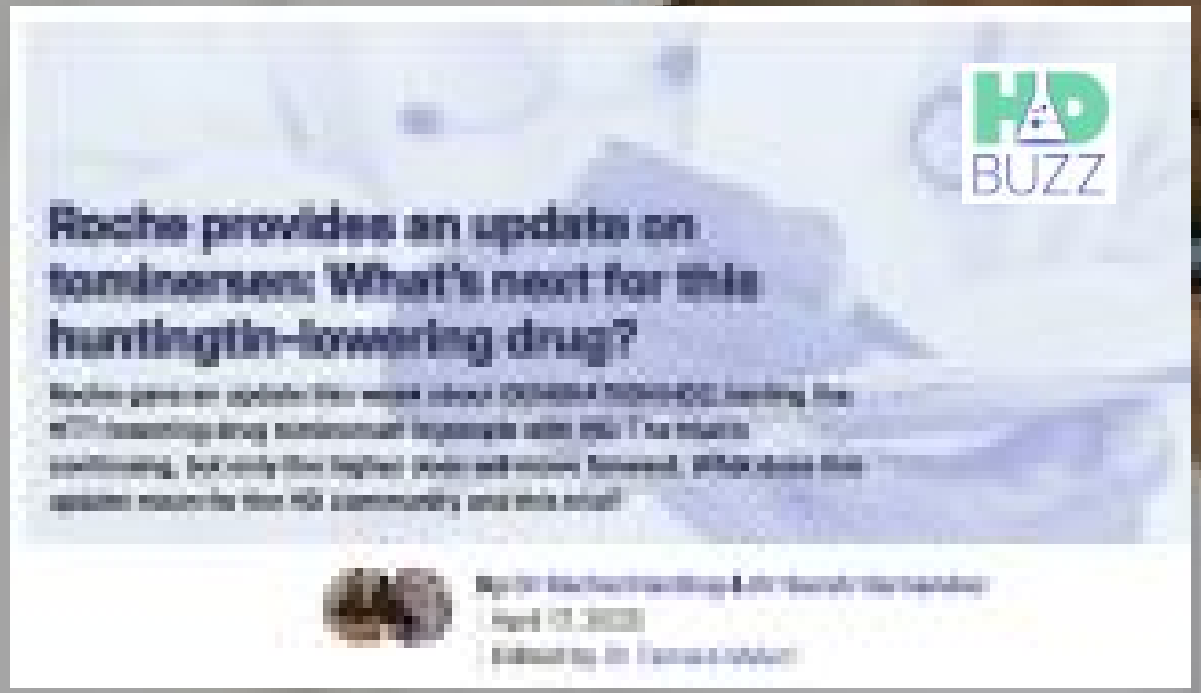


A Second Path: uniQure Plans Regulatory Filing for AMT-130 in the UK



uniQure

GENERATION-HD2
Trial for tominersen



An independent safety committee found no major safety concerns for the study and dosing is proceeding.

All participants will receive the 100 mg dose (or placebo) every 16 weeks - more likely to offer clinical benefit.

Data remains blinded

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SELECT-HD trial for WVE-003

- WVE-003 is safe and well tolerated
- Only lowers mHTT not wildtype HTT
- Some encouraging data from brain scans and other measures
- Next study in the works for trial with more people

VICO Therapeutics

HD BUZZ

Phase 1/2a clinical trial testing VO659

- VO659 reduces mutant huntingtin levels in people with HD
- It appears to be generally well-tolerated, but some participants developed radiculitis (nerve inflammation)
- Vico plans to adjust dosing to reduce side effects and extend dosing intervals
- A Phase 2 trial is being discussed for HD

NOVARTIS

PTC THERAPEUTICS

INVEST-HD
votoplam

POINT-HD showed that…

HD-ISS Stage 2: 10mg -> 52% slowing by cUHDRS; 5mg -> 28% slowing.

NfL remained below baseline for both doses

HD-ISS Stage 3: showed “potential signals” of benefit

Phase 3 INVEST-HD trial now launched, recruiting people with TFC=13 and early motor signs, a population where PIVOT-HD showed its strongest signal

Splice modulators could be working to lower HTT as well as targeting somatic instability

nature communications

Article

Splice modulators target *PMS1* to reduce somatic expansion of the Huntington's disease-associated CAG repeat

SKYHAWK THERAPEUTICS

Phase 2/3 FALCON-HD
SKY0515

SKY-0515 is safe and leads to dose-dependent lowering of huntingtin and PMS1

Targeting huntingtin and PMS1 could be a “two-for-one” effect on huntingtin lowering and somatic instability

Signals that cUHDRS might be improving for some participants

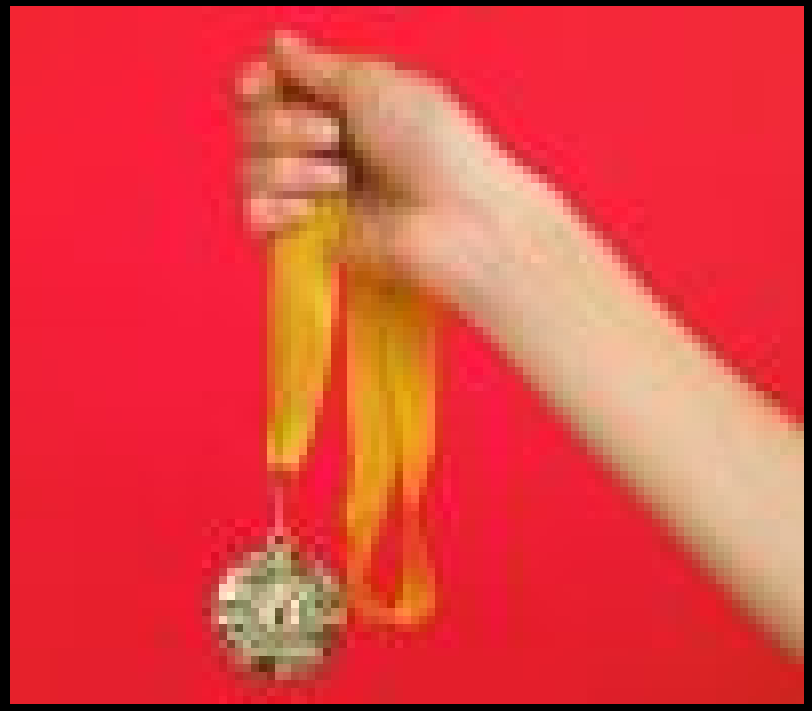
Phase 2/3 FALCON-HD trial is already underway but not recruiting in the US.

Provisional approval application in the works in Australia

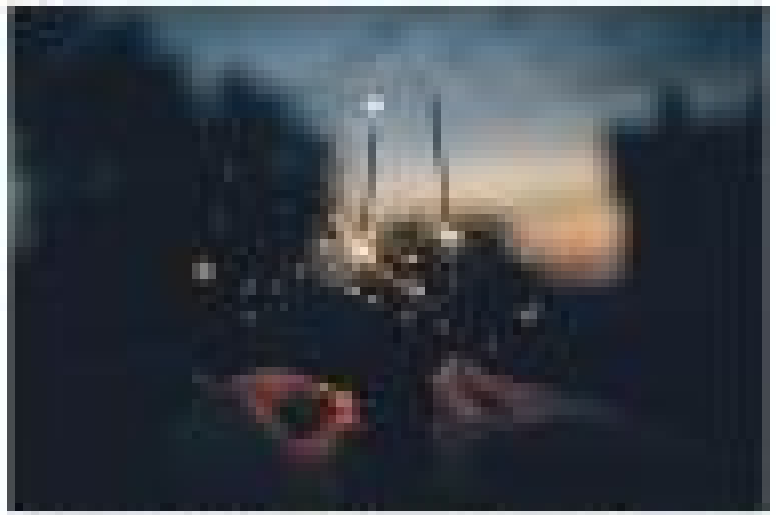
A lot of progress in ~10 years

Going boldly: First person treated in Phase 1 clinical trial by Alnylam Pharmaceuticals

A new Phase 1 clinical trial for the huntingtin-lowering drug ALN-HTT02 was initiated this week with the first dose given. Read on to learn details about the trial and how it compares to other ongoing huntingtin-lowering trials in the clinic.



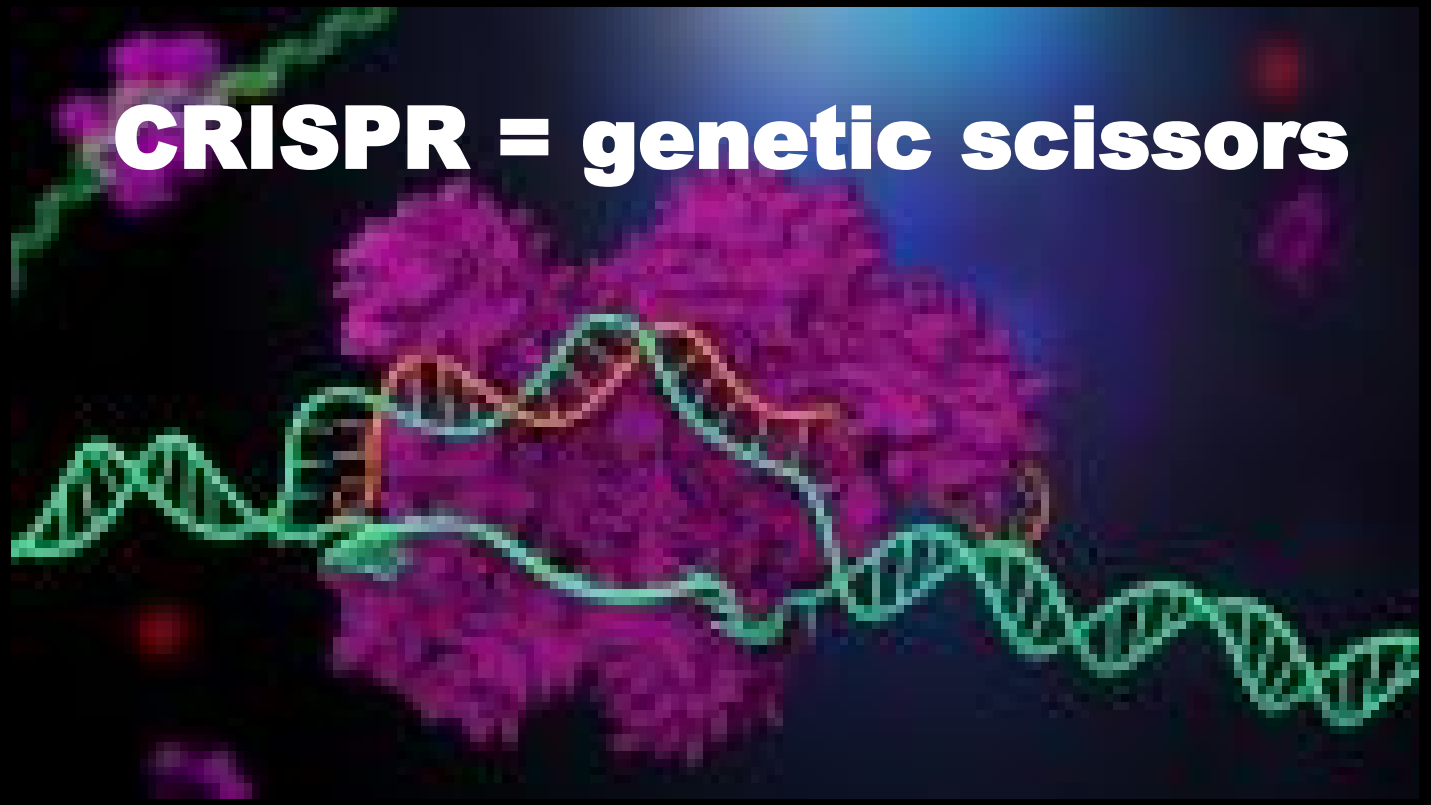
Lots of firsts in the HD clinical space!



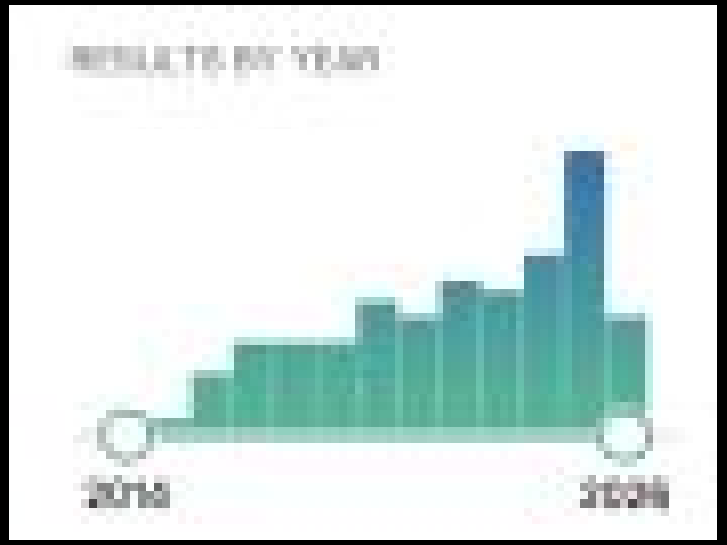
Spark ignited: first HD patient dosed in new Roche gene therapy trial

Spark and Roche issued a joint community letter to share that the first patient has been dosed in a new HD gene therapy trial.

CRISPR gene therapies can make permanent genetic changes to DNA



CRISPR could be used to reduce the CAG number, lower HTT, influence a genetic modifier….



Interest in using CRISPR in HD research is growing

CRISPR-based drugs: one giant leap for mankind

By Dr Sarah Hernandez | February 14, 2019

Edited by On Ranked Handling

You've likely heard of CRISPR. By now, you also that have heard that CRISPR has been used to produce a revolutionary new treatment for Sickle Cell Disease. Just 4 years after the Nobel Prize for the discovery of CRISPR was awarded, we have an approved treatment using this technology. This may have you wondering if this approach is being used in Huntington's disease in the research and when a similar drug for HD might come to the clinic. Lets discuss!

The genetic cause is known

The mechanism of disease is already known

Genetic editing can be done outside the body

Sickle Cell	HD
✓	✓
✓	✗
✓	✗

HDBUZZ

Permanent inactivation of Huntington's disease mutation by personalized allele-specific CRISPR/Cas9

Reversal of Phenotypic Abnormalities by CRISPR/Cas9-Mediated Gene Correction in Huntington Disease Patient-Derived Induced Pluripotent Stem Cells

Xiaohong Xu,[1] Yilin Tay,[2] Bernice Sim,[1] Su-In Yoon,[2] Yihui Huang,[1] Jolene Ooi,[1] Kagistia Hana Utami,[1] Amin Ziaei,[1] Bryan Ng,[1] Carola Radulescu,[1] Donovan Low,[1] Alvin Yu Jin Ng,[1] Marie Loh,[1] Byrappa Venkatesh,[2,3] Florent Ginhoux,[2] George J. Augustine,[2,4] and Mahmoud A. Pouladi[1,2,*]

CRISPR-Cas9 Mediated Gene-Silencing of the Mutant Huntingtin Gene in an In Vitro Model of Huntington's Disease

Silencing of the Mutant Huntingtin Gene through CRISPR-Cas9 Improves the Mitochondrial Biomarkers in an In Vitro Model of Huntington's Disease

CRISPR/Cas9-mediated gene editing ameliorates neurotoxicity in mouse model of Huntington's disease

CRISPR-Cas9-Mediated Genome Editing Increases Lifespan and Improves Motor Deficits in a Huntington's Disease Mouse Model

DNA double-strand break-free CRISPR interference delays Huntington's disease progression in mice

CRISPR/Cas9 Editing of the Mutant Huntingtin Allele In Vitro and In Vivo

Cutting to the chase with CRISPR

6 min read | A new study uses "self-switching" genetic scissors to target the root cause of Huntington's disease in mice – even after symptoms begin.

By Sonia Vázquez-Sánchez
April 16, 2026
Edited by Dr Leora Fox

CRISPR “molecular scissors” cut and disrupt the CAG expanded HTT gene directly in the brain of HD mice.

The CRISPR tool is self-inactivating, switching itself off after editing, improving safety.

Toxic protein levels reduced ~90%

Mice showed improved movement, behavior, and lifespan


Incisive
Genetics
HDBUZZ
life edit

Non-interventional studies

Registries

The global registry for Juvenile onset Huntington's Disease

Observational studies

Map the progression of the earliest HD related changes be found in young gene-positive adults

Surveys


HDYO
HDYO Survey Series
Share Your Voice to Help ALL
People Impacted by HD!

Global study with thousands of people

Yearly visits

Vast amounts of data
- How HD unfolds
- Tracking symptoms

Clinical trial design

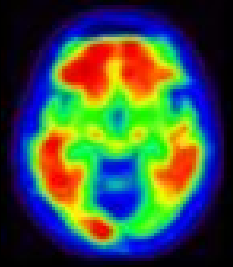
New targets

Observational research and local trials

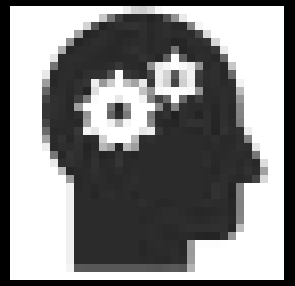
HDClarity

CSF Donation

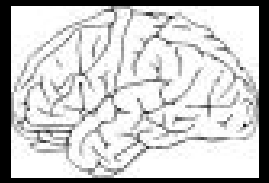
JHD & At-risk youth



Imaging studies



Cognitive testing



Brain Donation

Medication Combinations

Exercise and PT/OT

Huntington's Disease Society of America HDTrialfinder

Living an active and healthy lifestyle can help some HD symptoms

HDBUZZ

14 changes for a healthier brain

2024 HDBuzz Prize: Social Skills – The Hidden Gem in Improving Quality of Life for People with Huntington's disease?

Brain Gym: Staying Mentally Active May Slow Huntington's Disease

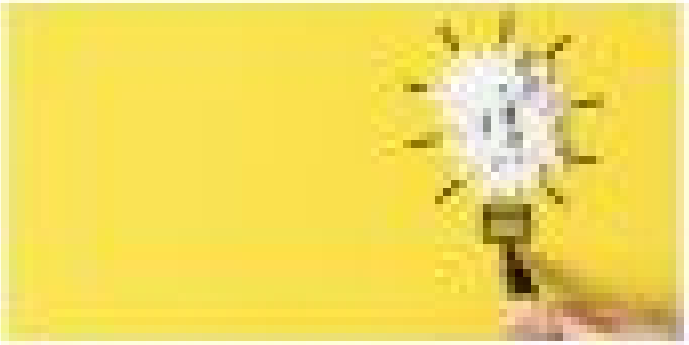
Wake up call: Sleep is impacted before Huntington's disease symptoms appear

HARVARD BRAIN TISSUE RESOURCE CENTER

UW Medicine
LABORATORY MEDICINE & PATHOLOGY

HDBUZZ

Landmark studies published made possible thanks to brain donations



New Insights Into Why Huntington's Disease Has Delayed Onset

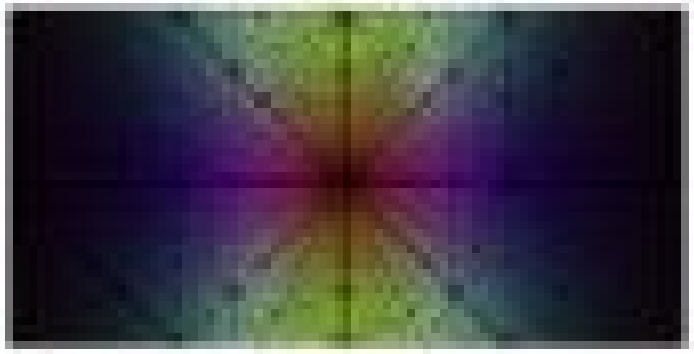
A highly-anticipated scientific paper has landed! This new work challenges current theories in Huntington's disease research, uncovering how runaway CAG repeats erode cell identity in certain types of brain cells, leading to their death.

Dr Sarah Hernandez January 28, 2023

Hats off to brain donors on Brain Donation Awareness Day

May 7 is Brain Donation Awareness Day. Today we highlight the selfless donation that many HD families have made, sending our gratitude, sharing research updates made with those precious brains, and detailing resources for brain donation.

Dr Sarah Hernandez May 07, 2023



Understanding expansions at the single cell level

Scientists have looked at CAG expansions in brains from people with HD to see which cells are affected

Dr Rachel Harding March 13, 2024

Article

Long somatic DNA-repeat expansion drives neurodegeneration in Huntington's disease

Robert E. Handsaker, Seva Kashin, Nora M. Reed, Steven Tan, Won-Seih Lee, Tara M. McDonald, Kelly Morris, Nolan Kamitaki, Christopher D. Mullally, Nadia R. Morakabati, Melissa Goldman, Gabriel Lind, Ross Knoll, Elizabeth Lawton, Marina Hogan, Kiku Ishihara, Sabina Berretta, and Steven A. McCarroll

Cell-type-specific CAG-repeat expansions and toxicity of mutant Huntingtin in human striatum and cerebellum

HDBUZZ

Huntington's Disease Society of America
HDTrialfinder

Huntington's Disease Foundation
Dedicated to Research

Huntington Society of Canada
HUNTINGTON
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HD
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