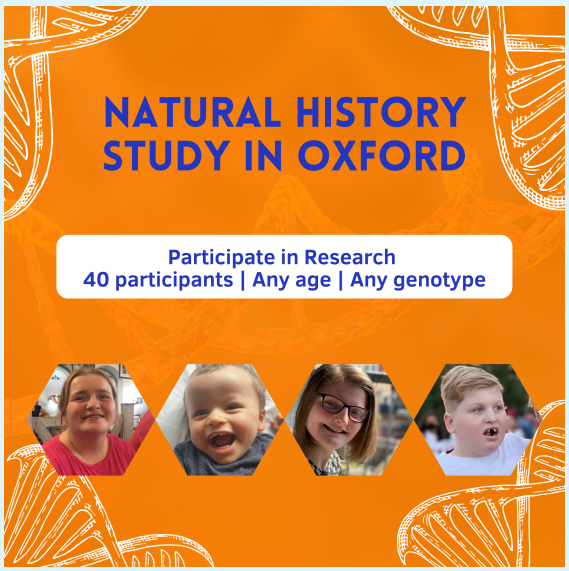
ANGELMAN SYNDROME THERAPEUTICS

Issue 1. August 2022



Recruiting for AS Natural History Study in Oxford

The Study is to assess and observe participants affected by Angelman syndrome (AS) over time with the aim to identify and design specific assessment tools tailored to the needs of Angelman syndrome patients.

If you know someone who lives with Angelman syndrome, please share this information with them and encourage them to participate.

Email theodora.markati@paediatrics.ox.ac.uk

The Study is invaluable for the development, trials, regulatory approval and NHS adoption of any future disease-modifying therapeutics for Angelman syndrome.

Active human clinical trials updates

GTX-102 by Ultragenyx 🇺🇸 🇨🇦 🇬🇧

Phase 1/2

Strategy: 'Unsilencing' of the paternal allele
Class: Antisense oligonucleotides (ASO)
Included genotypes: deletion

August 2022: Ultragenyx acquires GeneTx. The proceeds from the sale will be reinvested into AS research by FAST.
July 2022: No clinically significant safety issues at doses up to 10 mg with new administration strategy. Meaningful clinical activity in multiple domains on multiple measures. Enrollment of Cohorts 6 and 7 has begun: beginning at higher doses already cleared on safety.

Rugonersen by Roche 🇺🇸 🇮🇹 🇳🇱 🇪🇸

aka TANGELO trial

Phase 1

Strategy: 'Unsilencing' of the paternal allele
Class: Antisense oligonucleotides (ASO)
Included genotypes: deletion and mutation

August 2022: Final cohorts have now been enrolled. Long-term continuation program has been announced.

ION582 by Ionis 🇺🇸

aka HALOS trial

Phase 1/2

Strategy: 'Unsilencing' of the paternal allele
Class: Antisense oligonucleotides (ASO)
Included genotypes: deletion and mutation

August 2022: Multiple sites have been added, all in the US.
June 2022: ION582 receives Orphan drug and rare pediatric disease designations from FDA.

NNZ-2591 by Neuren 🇦🇺

Phase 2

Strategy: Downstream Treatments
Class: Agents to improve synapse growth, maintenance, and function
Included genotypes: all except mosaicism

July 2022: Phase 2 trial has started, in three clinics in Australia. Results from the trial are anticipated in H1 2023. In parallel with the trial, Neuren is also executing the foundational work to prepare for Phase 3 development.

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General information and contacts

What is Angelman syndrome?

Angelman syndrome is a rare neurogenetic disorder that affects approximately one in 15,000 people. It is estimated that ~500,000 individuals live with Angelman syndrome worldwide. Children and adults with Angelman Syndrome typically have severe cognitive and motor impairments, and can have debilitating seizures. Some individuals never walk. Most do not speak. Disruptive sleep also can be a serious challenge. All require 24/7 care throughout their lives.

How can I help?

It is estimated that about 4500 people live with Angelman syndrome in the UK. However, most of them are probably misdiagnosed with cerebral palsy, global developmental delay or autism due to the genetic testing being expensive and only becoming widely available in the last decade.

You can help by spreading the word about Angelman syndrome and the current state of research amongst your colleagues and with any professionals who treat or support people who live with Angelman syndrome. You can also encourage parents and carers of Angelman syndrome individuals to register in the Global Angelman Syndrome Registry.

Fundraising

Angelman Syndrome research in the UK does not receive government funding. Therrefore, all current research only has happended due to the fundraising efforts from parents and wider community. If you would like to support us, please donate or start your own Cure Angelman Now campaign - more details are on our website.

Upcoming events

Please join us virtually or in person for FAST 2022 Global Science Summit & Gala In Florida on 2-3 December 2022 to learn the latest scientific advances as well as pharmaceutical updates.

About us

FAST, the Foundation for Angelman Syndrome Therapeutics, was founded in 2008 in the USA with an urgent mission: cure Angelman Syndrome. Today, with operations also in Australia, Canada, France, Italy, Spain, LatAm and here in the UK, FAST has become the largest non-governmental funder of Angelman Syndrome research.

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