

TREATWolfram clinical trial to unlock a potential treatment for a rare disease

Birmingham is making a real-world impact on rare diseases, with a comprehensive multi-facility network for research helping to find new treatments for patients like those with Wolfram syndrome.



INTERVIEW WITH
Professor Tim Barrett
Professor of Paediatrics and Child Health in the Institute of Cancer and Genomic Sciences, Director of the Centre for Rare Disease Studies Birmingham and Chief investigator on the TREATWolfram trial

WRITTEN BY
Bethany Cooper

Rare Disease Day reminds us of the challenges faced by individuals grappling with rare disease. Though most of the population will not have a rare disease, it's vital that those who do receive equitable levels of care, research and treatment.

Getting a rare disease diagnosis

Shiffa, diagnosed with diabetes as a child, started insulin injections to manage her blood sugar. Vision symptoms emerged aged 6, leading to blindness. She says: "I had tests done in lots of places, and nobody could explain my symptoms."

For Shiffa, it took years to be diagnosed with Wolfram syndrome, a rare genetic disorder that affects only 1 in 500,000 people. Shiffa has now lost most of her sight, due to a gradual loss of brain cells, which may affect her hearing, balance and other vital functions. Alongside the physical symptoms, Shiffa stressed the importance of considering the emotional effects: "You have to talk to those around you. It is hard, but never give up."

Aiding Wolfram syndrome research

Wolfram Syndrome UK (WSUK), the UK charity aiding those impacted, was founded in 2010 by Paul and Tracy Lynch, after their daughter Jennifer's diagnosis. It initially supported five individuals. Now, it assists 120 — a figure rising due to better diagnosis.

Tracy acknowledges: "We have come a long way in the last 14 years. We're being recognised by larger organisations for our work supporting families affected by Wolfram syndrome, not just within the UK, but globally."

WSUK are currently supporting the incredible work being done by the University of Birmingham Cancer Research UK funded Clinical Trials Unit, which has pioneered the TREATWolfram international drug treatment trial. WSUK funds travel and lodging for participants and collaborates with specialist clinics in Birmingham while supporting global research efforts.

TREATWolfram trial brings hope

Shiffa was among the first patients recruited for the groundbreaking TREATWolfram trial. Researchers leading

the study have uncovered the disease mechanism and discovered a potential treatment. They're now spearheading the first global clinical trial of a repurposed drug for Wolfram syndrome.

The trial provides a beacon of hope for patients like Shiffa. Professor Tim Barrett, Chief investigator for the Wolfram trial, explains: "For patients with Wolfram syndrome, we hope to have treatments in the next few years which will slow or stop disease progression."

Birmingham as a rare disease hub

Shiffa relies on NHS England's Wolfram specialised service, delivered at University Hospitals Birmingham and Birmingham Women's and Children's Hospital, linked through Birmingham Health Partners. They host the UK's first adult and children's Clinical Centres for Rare Diseases, managing over 500 rare diseases and streamlining appointments in a 'one stop shop' model for those affected.

Birmingham Health Partners bring together the city hospitals and universities in an alliance to improve the health of the youngest and most diverse UK population outside London. Professor Barrett also identifies health inequality as the primary catalyst for Birmingham's rise as a rare disease research hub: "Birmingham's diverse population makes them disproportionately affected by rare diseases, while our location in the centre of the UK makes us accessible to patients and specialists from across the UK."

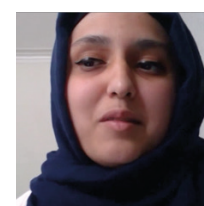
Leading rare disease treatment research

Barrett explains: "We are scaling up our trials to get more treatments to more patients with rare diseases, faster."

Birmingham's commitment to rare disease research encompasses the whole translational research pathway from discovery science through integrated phase trials to real world evidence of treatments in the clinic. Major collaborations with industry, University of Birmingham's world leading Cancer Research UK Clinical Trials Unit with specialised expertise in rare disease trials, and research supported by UK Medical Research Council and National Institute for Health Research make Birmingham the 'go to' city to recruit to and deliver translational research that will benefit our UK rare disease community.



INTERVIEW WITH
Tracy Lynch
Co-Founder & Chief Executive of Wolfram Syndrome UK Charity



INTERVIEW WITH
Shiffa Ahmed
Participant in the TREATWolfram Clinical Trial

Paid for by
University of Birmingham



Find out more at:
Birmingham.ac.uk/research/crcctu
Birmingham.ac.uk/research/rare-diseases

