
TREATING NONSENSE

A University College London researcher hopes to use a cystic fibrosis drug to cure forms of choroideremia

13 Dec 2016 by [Olivia Wannan](#)

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Cystic fibrosis and choroideremia patients have at least one thing in common – both have their lives significantly affected because they have a so-called ‘nonsense mutation.’ And a University College London (UCL) researcher hopes they will soon have another similar thing, a therapy in the form of the drug Ataluren.



‘Nonsense mutations’ mean that instead of a protein being created in its complete form, production stops prematurely, often leaving it functionless – with serious consequences for the body.

One in three choroideremia patients have this type of mutation, which affects the creation of the REP1 protein, UCL ocular biologist, Dr Mariya Moosajee, told *OT*.

To try to alleviate this problem, Dr Moosajee spent her PhD research trying to find a drug that weakens the effect of these ‘nonsense

mutations.' The ones she discovered were highly toxic, but then a new drug, Ataluren, was approved as a treatment for cystic fibrosis and Duchenne muscular dystrophy, she explained.

Using this drug on skin cells taken from a choroideremia patient, Dr Moosajee showed that production of complete and "healthy" REP1 was boosted, in [a paper](#) published in the journal *Human Molecular Genetics*.

The drug has a similar effect on zebrafish affected by a form of the ocular condition.

She emphasised that: "It's great when you see a therapy working...This has the potential to halt disease, or slow it down."

Following this Fight for Sight-funded research, Dr Moosajee is planning to launch a clinical trial offering Ataluren to suitable choroideremia patients in 18 months' time.

First, she will need to assemble a group of choroideremia patients with the 'nonsense mutations.' She is also preparing for the trial by analysing the best time during a patient's life to start prescribing such a drug.

Dr Moosajee highlighted that 'nonsense mutations' are also the cause of other serious ocular conditions, including type II Usher syndrome, adding: "This has the potential to work for many conditions."

Dr Moosajee is also working with pharmaceutical chemists to look at other drugs and compounds that work in a similar way to Ataluren.

Pictured is University College London researcher, Dr Mariya Moosajee (left), and research assistant, Dhani Tracey-White (right), are studying treatments for choroideremia.

Image credit: Mariya Moosajee/UCL

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