



Pioneering Usher syndrome research

We are immensely grateful for CUREUsher's ongoing support towards Professor Mariya Moosajee's research. She and her group are working hard to develop much-needed treatments for patients with Usher syndrome. Thanks to you, they will be able to drive their research further and faster.

Genes that cause some of the various types of Usher syndrome are very large. An example of this is USH2A, the gene most commonly responsible for Usher syndrome. Because the condition is primarily driven by larger genes, this has made developing suitable treatments more challenging.

Viral gene therapy – an approach which has been successful in treating other inherited conditions – can only deliver genes of a limited size (more than three times smaller than USH2A), meaning it is not a viable option for those living with Usher syndrome.

To overcome this, Professor Moosajee's team are developing a non-viral gene delivery system to encase USH2A. This alternative method has several benefits. It:

- Has the capacity to hold large genes.
- Does not integrate into the patient's DNA, thus reducing the risk of introducing cancer-related mutations.

- Does not have any viral components and therefore reduces the likelihood of a negative response from the body's immune system.
- Enables long-term gene expression in the body.

Together, this suggests that this approach is safe and effective for gene delivery.

Building on this, Professor Moosajee's team have shown the technique produces successful results in lab models which have Type 2 Usher syndrome caused by mutations in the USH2A gene, as well as in patients' cells. Her work is continuing to maximise the delivery of this novel gene therapy approach in larger models of the disease and assessing its safety.

Alongside this, Professor Moosajee's group are hoping to test a drug called ataluren in a small number of patients with USH2A caused by a specific type of mutation called a nonsense mutation. This will build the evidence they need to establish a full-scale clinical trial involving a much larger number of patients. You can read more about this approach on [Gene Vision's website](#).

Professor Moosajee and her team are hugely grateful for CUREUsher's continued support which is making all of this possible and powering their vital research to find new treatments for Usher syndrome. **Thank you.**