

Big challenges in research on Usher



The dream of people with Usher syndrome is that researchers will find in time a treatment to stop the process of becoming deaf and blind. Usher syndrome can't just be solved with 1 research or breakthrough. In this infographic we explain the obstacles finding a treatment for all people with Usher syndrome.

#stopUSH

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- 1 There are more than 10 Usher genes and some of them are way too large.



The existence of more than 10 Usher genes means that we are in need of different therapies to treat this disease. Some genes for Usher syndrome are way too large for the existing treatments to replace them. It is like trying to transfer big elephants sitting on a tiny Fiat Panda. The Usher genes can't be transferred into the eye and ear like that. Researchers are challenged to find alternative means of transport or to find ways to

- 2 There are many different gene mutations



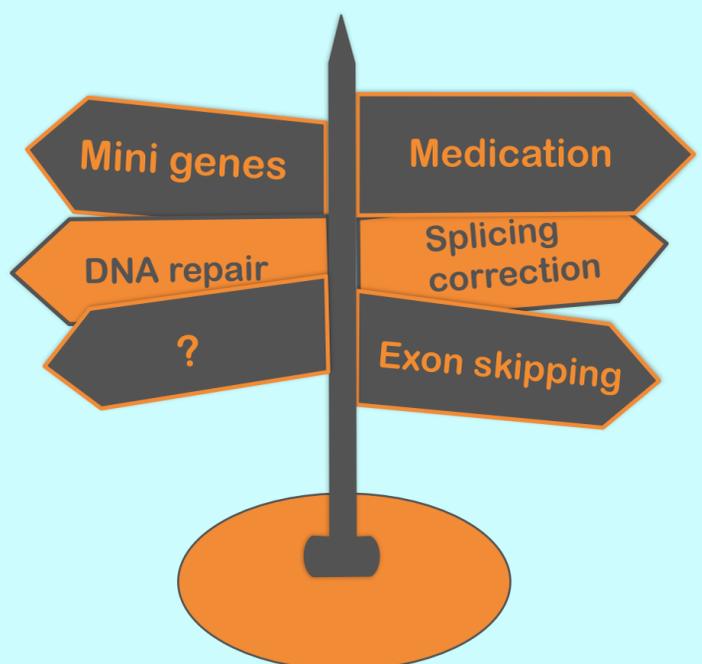
On these Usher genes there are more than 500 different mutations leading to the terrifying symptoms of Usher syndrome. Researchers need to develop alternative strategies for every specific mutation.

- 3 There is no appropriate model system for testing



Researchers need to work with cell lines and animal model in which Usher genes show their expression. They are almost not available. For example mice have barely problems with their sight despite of having these Usher genes. For now the zebrafish seems to be the best animal model which can become quantifiable deaf and blind.

The big challenge for researchers is that they need to head for a range of different research pathways to develop a treatment for all people with Usher syndrome. That is why Foundation Usher Syndrome stimulates all research lines, so that the dream to stop the process of deafblindness can be true for all people with Usher syndrome. #stopUSH!



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