

Expression of Interest in START eye drop therapy clinical trial for individuals with Aniridia due to PAX6 mutations

Dear Aniridia Network UK Conference participant,

You might have heard of the recent animal research on an exciting new eye drop therapy for aniridia. These eye drops are called START and contain the drug Ataluren. When the START eye drops were applied to the eyes of young mice with aniridia, some of the aniridia signs reversed.

Ataluren, the drug in START eye drops, works by skipping over certain types of gene alterations ('stop' mutations), meaning that the gene is restored to normal function. These are alterations that create an inappropriate early signal in the gene that says 'STOP', thereby preventing the synthesis of full-length gene product or protein. Ataluren makes the protein synthesis machinery in cells 'ignore' the STOP signal enabling the synthesis of the complete protein, restoring the protein back to normal levels. About 50% of individuals with aniridia have an alteration in the PAX6 gene that could be suitable for therapy using this type of treatment. The treatment has not been tried in humans before, but it is possible that some individuals could potentially benefit from START therapy, and this trial would evaluate this.

Prof. Cheryl Gregory-Evans, who led the study in mice, wishes to expand the work together with the manufacturers and start a clinical trial to find out if a similar reversal of aniridia symptoms will occur when aniridia patients with a nonsense mutation in the PAX6 gene are treated with the START eye drops. This trial will run from Vancouver, Canada.

The prospect of the possibility of therapy for congenital eye conditions, such as aniridia, would be very exciting. Currently there is no UK base for this trial, however, Professor Nicola Ragge, who works at Birmingham Women's Hospital and

Oxford Brookes University is very interested in setting up a clinic running this trial at the Institute for Translational Medicine, in Birmingham. Professor Ragge is a consultant geneticist and also a very experienced ophthalmologist who has a research team investigating congenital eye conditions at Oxford Brookes University. This work would be in collaboration with the group from Vancouver.

To be able to conduct a clinical trial with START eyedrops in the UK, Professor Ragge would need a certain number of participants, so she would like to know how many people are interested. If you are interested we would love to hear from you! You can contact us by leaving your details on the list that is provided in the venue, or you can call or email us directly.

We are looking forward to your replies!

Kind Regards,

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ANUK website on START therapy:

<http://aniridia.org.uk/2014/01/14/cure-for-aniridia-under-spotlight/>

Information about the drug:

<http://www.ptcbio.com/en/pipeline/ataluren-translarna/>