

MAC Research Update June 2019 from Dr Mariya Moosajee, MACS Medical Advisor

Dear Colleagues,

This has been an exciting year for our MAC research and I want to update you all about some of the projects my research group have been working on, including some opportunities for you to offer patients.



My name is Dr Mariya Moosajee, I am a Consultant Ophthalmologist at Moorfields Eye Hospital and Great Ormond Street Hospital for Children in London. I specialise in the genetics of developmental eye defects such as MAC. I also lead a research team at UCL Institute of Ophthalmology, London, where I am trying to understand the molecular mechanisms causing MAC and developing potential treatments that may be applicable in the future for patients and their families.

The NIHR Bioresource

The NIHR Bioresource is a national resource for researchers aiming to gain a better understanding of the relationship between the genetic cause and clinical features causing MAC with the aim to identify factors that influence disease severity- why some members of a family have a mild coloboma or unilateral features but others have bilateral microphthalmia. This research effort hopes to better understand the variation in the MAC spectrum, as well as develop new treatments.

We would like to encourage patients to be a part of this valuable resource. Everyone with MAC is eligible, and would need to provide clinical information and blood samples.

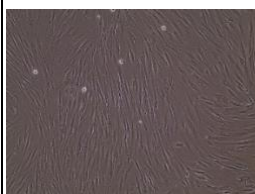
For more information, go to:

<https://bioresource.nihr.ac.uk/wp-content/uploads/2018/06/Web-summary-MAC-v1.1-20052019.pdf>

Growing models of microphthalmia in the lab

Using cells from a small piece of a patient's skin, my research team are able to generate stem cells. By adding nutrients, we encourage these cells to grow into early eye-cups, which can be used to model eye development. We are now looking to use stem cells derived from patients with MAC to grow these early eyes in a dish.

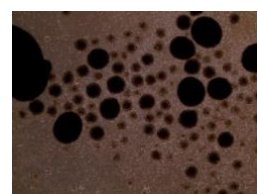
Skin Cells



Stem Cells



Early Eye-Cups



We hope to use these models to understand the genetic pathways involved in eye development associated with MAC, as well as test new drugs.

To learn more, watch our video - seeing with your skin:

<https://vimeo.com/227432768>

To keep track of our research, please follow:

@MariyaMoosajee on Twitter

DrMariyaMoosajeeLab on Facebook

Please contact us if you need genetic testing for patients or have any questions relating to this research. We will be happy to help you!

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