Moorfields Eye Hospital NHS
NHS Foundation Trust

PRESS RELEASE

£1.1 million funding awarded to tackle child blindness

A Moorfields consultant has received a major funding award to identify the genetic causes and develop targeted gene therapies to prevent the commonest cause of blindness in children.

Dr Mariya Moosajee, consultant ophthalmologist at Moorfields Eye Hospital NHS Foundation Trust and senior clinical lecturer at UCL Institute of Ophthalmology has been awarded £1.1m from the Wellcome Trust for a Clinical Research Career Development Fellowship.

Stem cells derived from patient's skin will be used to grow a 3D model of a human eye to investigate genes that cause ocular maldevelopment, a condition which can cause babies to be born without eyes or underdeveloped eyes. Pioneering gene editing technology will then correct the defective gene to see if the cells can grow into normal healthy eyes.

It is hoped that this could pave the way for targeted drug and gene therapies that might one day be used as a prenatal therapy. Birth eye defects develop during pregnancy and cause structural abnormalities in the eye. They are responsible for over a third of blindness and severe visual impairment in children worldwide. Only a few genes have been found to cause the abnormalities and there is no treatment currently available.

The funding will allow for full genome screening of patients with birth eye defects to identify the genetic cause of their condition. This will help provide accurate genetic diagnosis in the future, with informed genetic counselling and improved care pathways for patients and their families.

Dr Mariya Moosajee, consultant ophthalmologist at Moorfields Eye Hospital, said: "Nearly twenty percent of all children registered blind in the UK suffer from birth eye defects but only a few genetic causes have so far been identified. Once we understand the genetic causes and can identify chemical changes in human DNA that influence genes being switched on or off at crucial points during normal eye development, a real focus on developing a treatment can be made. It will allow us to develop and test new drugs and targeted genetic therapies that could one day be used to encourage eye growth and prevent the abnormalities from developing."

Professor Sir Peng Tee Khaw, director of research and development at Moorfields Eye Hospital and director of the NIHR Biomedical Research Centre (BRC) at Moorfields and the UCL Institute of Ophthalmology, said: "This is an extremely important and exciting award from the Wellcome Trust to Dr Moosajee. So many of the children we see here at the NIHR Biomedical Research Centre at Moorfields and the UCL Institute of Ophthalmology, where we have the largest children's eye centre in the world, suffer from these developmental problems. This research will help us to translate this

leading science into new understanding and hopefully treatments for these children in the future."

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Dr Mariya Moosajee is available for interview on request. Please contact the press office on 020 7566 2628 for further information.

For further information, please contact the Moorfields Eye Hospital Press Office on press.office@moorfields.nhs.uk or call **020 7566 2628**. For out of hours enquiries please call **020 7253 3411** who will contact the on call press officer.

Notes to editors:

Ocular maldevelopment (birth eye defects), cause structural defects in the eye which occur within the first four to eight weeks of pregnancy. The defects include: microphthalmia, a disorder in which one or both eyes are abnormally small; anophthalmia, the absence of one or both eyes; coloboma, cleft of the eye.

Birth eye defects are seen in 10 per 100,000 births and cause over a third of severe visual impairment and blindness in children worldwide.

It is the most common cause of childhood sight impairment certification in England and Wales, accounting for 18.4%. In 2013, the Royal National Institute for the Blind (RNIB) reported that 25,000 children aged between 0–16 years were registered blind or visually impaired in the UK.

The James Lind Alliance and Sight and Vision Priority Setting Partnership lists the causes and prevention of ocular maldevelopment within the top ten research priorities into childhood onset disorders.

About Moorfields:

- Moorfields is one of the leading providers of eye health services in the UK and a world class centre of excellence for ophthalmic research and education. Our main focus is the treatment and care of NHS patients with a wide range of eye problems, from common complaints to rare conditions that require treatment not available elsewhere in the UK. Our unique patient case-mix and the number of people we treat mean that our clinicians have expertise in discrete ophthalmic sub-specialties.

- In 2015/16 we saw more than half a million patients in our outpatient services and carried out almost 40,000 surgical procedures, making Moorfields the largest ophthalmic provider in the UK. We also provided care to 104,000 patients in our A&E department.

- We treat people in 32 locations in and around London, the south east and Bedford, enabling us to provide expert treatment closer to patients' homes. We also operate commercial divisions that provide care to private patients in both London and the Middle East.

- Moorfields' innovative approach to delivering care across multiple satellite sites has been explicitly referenced in recent national policy. The *Five Year Forward View*

highlighted the benefits of our model in helping to sustain local hospital services and enable smaller hospitals to remain viable. The Dalton Review categorised our approach as a contractual arrangement which it described as a service-level chain. More recently, the Moorfields@ model has been cited as an example of franchising or networked care. Terminology will be important as we seek to describe the models that could be replicated across the NHS. In this value proposition we use the term 'networked care' to describe the generic model of collaboration between providers and the term 'satellite model' to describe the approach currently delivered by Moorfields.

- With our academic partners at the UCL Institute of Ophthalmology, Moorfields is recognised as a leading centre of excellence in eye and vision research. Together we form one of the largest ophthalmic research sites in the world, with the largest patient population in Europe or the USA. We publish more scientific papers than any other eye and vision research site and have an extensive joint research portfolio.

National Institute for Health Research (NIHR) Biomedical Research Centre at Moorfields Eye Hospital NHS Foundation Trust and UCL Institute of Ophthalmology was established in April 2007 and has recently been awarded a third five-year term by the NIHR from April 2017. Its purpose is to conduct 'translational research' that is designed to take advances in basic medical research from the laboratory to the clinic, enabling patients to benefit more quickly from new scientific breakthroughs. Our centre is currently one of 20 biomedical research centres that have been awarded from April 2017 to NHS/university partnerships with an outstanding international reputation for medical research and expertise, and experience of translating that research into the clinical setting. For further information, please visit www.brcophthalmology.org.

UCL Institute of Ophthalmology is one of a number of specialised research centres within UCL (University College London) and is, together with Moorfields Eye Hospital, one of the leading centres for eye research worldwide. The combination of the Institute's research resource with the resources of Moorfields Eye Hospital, which has the largest ophthalmic patient population in the Western World, opens the way for advances at the forefront of vision research. For further information, please visit www.ucl.ac.uk.

About Wellcome:

Wellcome exists to improve health for everyone by helping great ideas to thrive. We're a global charitable foundation, both politically and financially independent. We support scientists and researchers, take on big problems, fuel imaginations and spark debate.