

# Genetic mutation identified for eye complaint

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An international research collaboration including research teams from the Children's Hospital in Boston (USA), King's College London and the Peninsula Medical School, has identified a gene that, when mutated, causes Duane syndrome. The research is published in the latest edition of *Science*.

Duane syndrome is a congenital eye movement disorder that causes eye muscles to contract and relax when they should not. It develops in the womb where it affects nerve growth in the eye. Sufferers of the syndrome have limited eye movement sideways towards the ear or nose. When the eye moves towards the nose the eyeball pulls into the socket, closing the eyelids and sometimes forcing the eye movement up or down.

Duane syndrome usually causes vision problems. It can be accompanied by malformations of the skeleton, eyes, ears, kidneys and nervous system but more commonly occurs in isolation.

Most sufferers of the condition are diagnosed by the age of 10. It is more prevalent in women and in the left eye. It is believed to affect around half a million people worldwide.

Until now, scientists have been unable to pinpoint a genetic mutation that causes isolated Duane syndrome. By isolating the gene from affected families, the research team introduced the mutation into chick embryos (which have a visual system very similar to that of humans) and

produced similar defects in nerve growth.

Dr. John Chilton, RCUK Academic Fellow in Clinical Neuroscience and Molecular Biology at the Peninsula Medical School, commented: "By understanding how this gene causes Duane syndrome, we can begin to achieve a wider understanding of how the visual system develops in the womb. This raises the possibility of better diagnosis and even genetic treatments for visual conditions such as Duane syndrome'."

He added: "We also discovered that the gene responsible for Duane syndrome is widely expressed throughout the nervous system, so the next question to be answered is why only the nerves that control the eye muscles are affected."

The international research collaboration includes Dr. Elizabeth Engle at the Children's Hospital in Boston, USA; Dr. Chilton at the Peninsula Medical School (PMS); Dr. Nick Gutowski, Professor Sian Ellard and Dr. Beth Young at both PMS and the Royal Devon and Exeter Hospital; Professor Sarah Guthrie at King's College London.

## CASE STUDY

Ami Ryder, aged 8 from West Park, Plymouth, UK, was diagnosed with Duane syndrome as a small baby at the Royal Eye Infirmary, Plymouth Hospitals NHS Trust.

Since then Ami, who is a pupil at Knowle Primary School, has undergone a series of treatments at the Royal Eye Infirmary for type B Duane syndrome in her left eye. Although Ami does not have a full range of eye movement, she turns her head to maintain vision and it has not stopped her enjoying skipping and football.

Her doctors say she is doing very well and, at age 8, her vision is not

likely to deteriorate any further.

Her mum, Sarah Ryder, said: "Ami is great, she just gets on with it and thinks it is actually quite funny. It certainly hasn't stopped her enjoying all the activities that every little girl of her age enjoys."

Sarah continued: "We are very pleased with her treatment and we are delighted to have been told that there will be no lasting damage to her eyesight. She has taken the regular visits to hospital in her stride, and she has now been told that all she needs is one final examination and that's it."

Lizzy Padgett, Head Orthoptist at the Royal Eye Infirmary, commented: "Although the eye muscle balance of patient's with Duane syndrome is affected in such a way that the patient does not have the full range of eye movement, the majority of patients are in fact often asymptomatic and control any squint by turning their head to maintain binocular vision. The concern with children who have a squint due to Duane syndrome is the risk of developing amblyopia (where the brain favours one eye over the other), but this is usually treatable during childhood."

She added: "The breakthrough made by colleagues at the Peninsula Medical School and their research partners in the US and UK is extremely welcome and should lead to more advances in our knowledge of how the eye and visual system develops."

Source: The Peninsula College of Medicine and Dentistry

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