

Genetic clue to common birth defects found

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Scientists at King's College London have, for the first time, uncovered a gene responsible for Adams-Oliver Syndrome (AOS), a condition which can cause birth defects of the heart, limbs, or blood vessels.

The study, published in The [American Journal of Human Genetics](#) today, gives valuable [insight](#) not only into this particular condition, but also the possible genetic causes of these common [birth defects](#) found in the wider [population](#).

The team of researchers, led by the National Institute for Health Research (NIHR) comprehensive Biomedical Research Centre (BRC) at King's College London and Guy's and St Thomas', say that these findings could lead to better ways of treating children with these defects and may, in the future, help to find ways to recognise and ultimately prevent them from occurring.

AOS is a rare developmental condition that affects less than 150 families worldwide. But birth defects of the heart, limbs, and [blood vessels](#), seen in [babies](#) with the condition, are in fact relatively common in the general population – for example, 9 in every 1,000 babies are born with a heart defect.

The team of researchers set out to investigate the genetic cause of AOS in order to detect clues to the role genes might play in congenital birth defects.

Using modern DNA technology to examine the patterns and variation of

genes within two affected AOS families, the team detected mutations in the ARHGAP31 gene. This gene regulates two proteins in the body with important roles in cell division, growth, and movement. Mutations in the gene result in an imbalance in the regulation of these proteins, most likely leading to a disruption of the signalling proteins that are critical for normal limb formation.

Professor Richard Trembath, Head of King's College London's Division of Genetics and Molecular Medicine and Medicine Director of the NIHR BRC, said, "Birth defects of the [heart](#), limbs and blood vessels can cause distress for children and their families, and tragically can sometimes even be fatal."

"Through this study we have uncovered the first inherited factor associated with Adams-Oliver Syndrome, which gives us greater understanding of how associated birth defects develop. Understanding the genetic causes of rare diseases in this way not only helps us to understand the condition better, but it gives us a unique insight into the role of specific genes in human development on a broader scale."

"Ultimately, this knowledge may lead us to develop better ways of treating children with these kind of abnormalities, and one day we may even be able to prevent them from developing in the first place."

Provided by King's College London

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