

Study discovers link in childhood brain disease research

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University of Manchester scientists at the National Institute for Health Research Manchester Biomedical Research Centre (BRC) have discovered a new link between a rare childhood disorder and a common immune system disease.

Over the last 10 years Professor Yanick Crow and an international team of colleagues have studied the inflammatory [brain disease](#) Aicardi-Goutières syndrome (AGS). Some children with AGS also develop an early-onset form of the autoimmune disorder systemic lupus erythematosus (SLE).

The researchers previously discovered that mutations in four genes caused AGS. These genes provide the instructions for making nucleases, a type of protein which cleans up 'waste' nucleic acids produced during the normal life-cycle of our cells. A failure of this clean-up process causes waste DNA and other material to accumulate in cells. The immune system then mistakes this material as 'foreign' and attacks it.

Professor Crow, based in the School of Clinical and Laboratory Sciences, said: "We have now identified mutations in a fifth gene, accounting for approximately 20% of AGS cases. This result is important because it will allow for confirmatory genetic testing in children with suspected AGS, and the opportunity of genetic testing during pregnancy for affected families. Additionally, our data adds a previously unknown component to the puzzle of how the innate immune response is triggered in AGS and lupus.

"Further studies of this gene will increase our understanding of the precise mechanisms involved in causing AGS and SLE, and thus allow us to develop precisely targeted therapies for these devastating disorders. This is work that is ongoing in our BRC-supported laboratory."

The importance of the discovery linking the causes of AGS and SLE has led to the team's findings being published in the leading international magazine *Nature Genetics*.

Source: University of Manchester ([news](#) : [web](#))

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