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News Release

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Cell study sheds light on diseases caused by immune system fault

Scientists have discovered how a gene mutation can lead to diseases that occur when the immune system attacks the body by mistake.

Understanding how these mechanisms work could help scientists to develop new treatments for autoimmune diseases such as Lupus and neurodegenerative conditions including Motor Neurone Disease.

Researchers found that a mutation in the gene – called *ADARI* – causes a defect in an alarm system in our cells that normally protects the body from viruses and other infections. This means that the alarm system is tripped by the cell's own molecules, causing the immune system to attack.

Some viruses encode their genetic material using a molecule called RNA, which is also produced by our own cells when the genes encoded in our DNA are translated into proteins.

Detecting foreign RNA inside a cell – such as from a virus – is the first warning sign of an infection.

Researchers – led by the University of Edinburgh – have uncovered a system that helps the body to differentiate between normal RNA and RNA from foreign organisms.

They found that ADAR1 adds a distinct chemical signature to the cell's own RNA molecules to stop them from setting off an immune response. Virus RNA molecules lack this and so are detected by the cell's alarm system.

Defects in the cell's ability to modify its RNA could lead to the triggering of immune responses against the cell itself when there is no infection present, the scientists say.

Mutations in the *ADARI* gene have been linked to a rare autoimmune disease called Aicardi-Goutieres Syndrome. The disease affects the brain and skin and most people who are affected have significant intellectual and physical problems.

The syndrome usually takes hold in early childhood and is so rare that it is not known how many people are affected. However the disease shows significant resemblances to the much more common and less severe Lupus Disease.

Ranked among the top universities in the world

The study is published in the journal *Cell Reports* and was funded by the Medical Research Council.

Dr Liam Keegan, of the MRC Human Genetics Unit at the University of Edinburgh, said: “Our findings provide fresh insights into the way cells distinguish our own genetic material from that of disease-causing agents, such as viruses.”

Dr Mary O'Connell, of the MRC Human Genetics Unit at the University of Edinburgh, said: “This is crucial for understanding how the immune response can be inappropriately triggered to attack our own cells and cause devastating autoimmune and neurodegenerative diseases.”

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