

Uncovered: Genetic cause of complex disease seen in Irish Traveller community

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Two independent groups of researchers — one led by Adrian Clark, at Queen Mary University of London, United Kingdom; and the other led by Jean-Laurent Casanova, at The Rockefeller University, New York — have now identified the disease-causing gene in patients with a complex inherited syndrome most commonly observed in the Irish Traveller community.

As noted by Jordan Orange, at the University of Pennsylvania School of Medicine, Philadelphia, in an accompanying commentary, the new data provide deep mechanistic insight into a complex human condition and expand our understanding of the human immune and endocrine systems, both of which are disrupted in patients.

Within the Irish Traveller community, several families have been found to suffer from an inherited condition characterized by failure of the [adrenal glands](#) to produce adequate amounts of steroid hormones, abnormal development (in particular, retarded growth), and a deficiency in immune cells known as NK cells. Both groups of researchers found that mutations in the MCM4 gene are responsible for this complex inherited condition. The MCM4 gene is responsible for templating a protein that is required for DNA to replicate itself, something that happens every time a cell divides.

Consistent with this, both groups of researchers found that the MCM4 mutations associated with disease caused genomic instability, something that they suggest might possibly put affected individuals at increased risk

for cancer.

More information: MCM4 mutation causes adrenal failure, short stature, and natural killer cell deficiency in humans. View this article at: [www.jci.org/articles/view/6022 ... cb4664b3464dffe5841c](http://www.jci.org/articles/view/6022...cb4664b3464dffe5841c)

Partial MCM4 deficiency in patients with growth retardation, adrenal insufficiency, and natural killer cell deficiency. View this article at: [www.jci.org/articles/view/6101 ... 9c3e63bab008bc523ccd](http://www.jci.org/articles/view/6101...9c3e63bab008bc523ccd)

Unraveling human natural killer cell deficiency, View this article at: [www.jci.org/articles/view/6262 ... 7b2cf2771a25813f75d8](http://www.jci.org/articles/view/6262...7b2cf2771a25813f75d8)

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