

## Tiny genetic alteration linked to devastatingly large brain size

An international collaboration led by scientists and doctors from the UK, Netherlands and United States has identified a new genetic disease that affects the size of our brains and causes severe developmental problems. While working on another project, Dr Siddharth Banka who is based at The University of Manchester and the [Manchester Centre for Genomic Medicine](#), Saint Mary's Hospital, noticed three children with large or small brains, developmental delay and mutations in a gene called RAC1.

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Professor Han Brunner and Dr Margot Reijnders of the Radboud University in Nijmegen had gathered together four more children with mutations in the same gene and altered brain-sizes. Some of the affected children were also found to have epilepsy and heart defects.

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“[O]ccasionally a tiny little alteration in just one of these thousands of genes can have devastating effects, reflecting the fine balance of this complex genetic orchestra.” [said Dr. Banka]

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“We suspect RAC1 plays an important role in other genetic diseases as well”. Dr Banka added: “We are sure that this discovery will lead to identification of more patients with RAC1 mutations, who have not yet been diagnosed. “The next step for the scientists will be to use this knowledge to work out possible treatments for this disease and related conditions.”

**The GLP aggregated and excerpted this blog/article to reflect the diversity of news, opinion, and analysis. Read full, original post: [Devastating disease which changes brain size discovered](#)**