Still-unnamed rare genetic disease linked to abnormal brain development in children

Scientists have discovered a new genetic disease, which causes some children's brains to develop abnormally, resulting in delayed intellectual development and often early onset cataracts.

The majority of patients with the condition, which is so new it doesn't have a name yet, were also microcephalic, a birth defect where a baby's head is smaller than expected when compared to babies of the same sex and age.

Researchers from the universities of Portsmouth and Southampton found that changes in a gene called coat protein complex 1 (COPB1) caused this rare genetic disease.

Now the variant has been identified, it will help clinicians come up with targeted interventions to help patients and their families, also opening the door to screening and prenatal diagnosis.

Co-author Diana Baralle, Professor of Genomic medicine and a clinical geneticist at the University of Southampton, said: "Next generation sequencing is transforming our ability to make new diagnoses and discover new causes for rare disorders. This story started with sisters I saw in clinic without a known underlying cause for their signs and symptoms. Looking closely at their genes, along with further functional molecular work and xenopus studies, we saw that this was a new previously undescribed syndrome. A diagnosis is so important to the family."

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