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Researchers find multisystem disorder caused by CCDC47 variants

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Researchers and clinicians through a multicenter collaboration have identified a novel multisystem disorder caused by bi-allelic variants in the CCDC47 gene. Their findings are reported in the *American Journal of Human Genetics*.

CCDC47 is responsible for encoding an essential calcium (Ca2+)-binding protein involved in embryogenesis and development. Calcium signaling is essential for various cellular processes including muscle contraction, secretion regulation, cell proliferation, and gene transcription.

In this study, detailed clinical characterization and functional studies were performed on four unrelated individuals with a complex multisystem disorder characterized by woolly hair, liver dysfunction, itchy skin, unusual facial features, low muscle tone, and global developmental delay.

Whole exome sequencing and family-based genomic analyses were performed at three different research centers and were used to identify the underlying genetic variants in CCDC47 associated with the condition. In this study, cells from individuals with the damaging alleles showed decreased CCDC47 gene expression and protein levels, supporting the pathogenic nature of these variants.

Two of the four patients included in this study were evaluated at The Community Health Clinic in Topeka, IN by clinicians from the Clinic

for Special Children and The Community Health Clinic.

The study demonstrates the important role of CCDC47 in normal development and the clinical consequence of its absence. The authors note that the identification and characterization of additional individuals will help further clarify key clinical features of this multisystem disorder, as patients demonstrated some variability in the clinical presentation of the disorder.

More information: Marie Morimoto et al, Bi-allelic CCDC47 Variants Cause a Disorder Characterized by Woolly Hair, Liver Dysfunction, Dysmorphic Features, and Global Developmental Delay, *The American Journal of Human Genetics* (2018). DOI: 10.1016/j.ajhg.2018.09.014

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