



**Clinic for  
Special Children**



**The Community  
Health Clinic**

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**FOR IMMEDIATE RELEASE**

## **RESEARCHERS FIND MULTISYSTEM DISORDER CAUSED BY *CCDC47* VARIANTS**

STRASBURG, PA & TOPEKA, IN- 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 (Ca<sup>2+</sup>)-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.



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The research was conducted by a team including the study's co-lead authors Marie Morimoto,<sup>1,15</sup> Helen Waller-Evans,<sup>2,15</sup> Zineb Ammous,<sup>3,15</sup> Xiaofei Song,<sup>4,15</sup> Kevin A. Strauss,<sup>5</sup> Davut Pehlivan,<sup>4</sup> Claudia Gonzaga-Jauregui,<sup>6</sup> Erik G. Puffenberger,<sup>5</sup> Charles R. Holst,<sup>7</sup> Ender Karaca,<sup>4</sup> Karlla W. Brigatti,<sup>5</sup> Emily Maguire,<sup>2</sup> Zeynep H. Coban-Akdemir,<sup>4</sup> Akiko Amagata,<sup>7</sup> Christopher Lau,<sup>1</sup> Xenia Chepa-Lotrea,<sup>1</sup> Ellen Macnamara,<sup>1</sup> Tulay Tos,<sup>8</sup> Sedat Isikay,<sup>9</sup> Michele Nehrebecky,<sup>1</sup> John D. Overton,<sup>6</sup> Matthew Klein,<sup>7</sup> Thomas C. Markello,<sup>1</sup> Jennifer E. Posey,<sup>4</sup> David R. Adams,<sup>1,10,11</sup> Emyr Lloyd-Evans,<sup>2</sup> James R. Lupski,<sup>4,12,13,14</sup> William A. Gahl,<sup>1,10,11</sup> May Christine V. Malicdan<sup>1,10,11</sup>

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### **About the Clinic for Special Children**

The Clinic for Special Children (CSC) is a non-profit organization located in Strasburg, PA, which provides primary pediatric care and advanced laboratory services to those who suffer from genetic or other complex medical disorders. Founded in 1989, the organization provides services to over 1,050 active patients and is recognized as a world-leader in translational and precision medicine. The organization is primarily supported through community fundraising events and donations. For more information, please visit [www.ClinicforSpecialChildren.org](http://www.ClinicforSpecialChildren.org)

### **About The Community Health Clinic**



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**The Community  
Health Clinic**

The Community Health Clinic (CHC) is a non-profit organization healthcare facility located in Topeka, IN, which provides clinical genetic services to all individuals, regardless of religion, race, or age. Services include, but are not limited to, access to a clinical geneticist, physician assistant, nurse, metabolic dietician, and genetic counselor. The CHC embraces, incorporates and promotes participation in research to advance medical knowledge and improve care. For more information, please visit <http://indianachc.org/>