



CENTOGENE, University College London, and Global Team of Researchers Discover Gene Associated With New Neurodevelopmental Disease Linked to Early-Onset Dystonia and Parkinsonism

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Results From Landmark Study Published in Brain Journal

- Collaborative research initiative leveraged CENTOGENE's Whole Exome Sequencing (WES) to reveal disease-causing gene called *ACBD6* (Acyl-CoA Binding Domain Containing 6)
- Over seven years, a total of 45 affected individuals from 29 independent families in Southeast Asia, Central Asia, the Middle East, Europe, and North and South America have been identified
- Study guides further research for potential treatments, including genetic causes and pathways leading to Parkinson's disease

CAMBRIDGE, Mass. and ROSTOCK, Germany and BERLIN, Nov. 14, 2023 (GLOBE NEWSWIRE) -- Centogene N.V. (Nasdaq: CNTG), the essential life science partner for data-driven answers in rare and neurodegenerative diseases, today announced the discovery of a new form of early-onset dystonia and parkinsonism in the context of neurodevelopmental abnormalities associated to the gene called *ACBD6* (Acyl-CoA Binding Domain Containing 6) as part of an international team of researchers. The landmark study's findings have been published in [Brain](#), a leading, peer-reviewed scientific journal of neurology.

The study reports on 45 patients from 29 families and provides evidence that bi-allelic pathogenic variants in the *ACBD6* gene lead to a distinct neurodevelopmental syndrome accompanied by complex and progressive cognitive and movement disorders. The clinical features were similar across all patients, and the study identified a recognizable clinical pattern that will help clinicians diagnose this disease.

"Over the last years, CENTOGENE played a key role in identifying relevant patients using Whole Exome Sequencing (WES) and providing genomic and phenotypic analysis as well as overall mapping of clinical data," said one of the paper's lead authors, Dr. Aida Bertoli-Avella, Head of Research Data Analysis at CENTOGENE. "The findings have helped us gain a deeper understanding of neurological disorders – offering a next step towards advancing treatments and life-changing answers for these patients, their families, and the greater neurological disease community."

To enhance their understanding of the disease's mechanism, the research team has embarked on generating models that might help identify potential therapeutic targets. As the disease is associated with parkinsonism, a better understanding of the function of the *ACBD6* gene through further studies could also shed light on genetic causes and pathways leading to Parkinson's disease.

Dr. Reza Maroofian from the University College London Queen Square Institute of Neurology who led the project said, "This study reinforces the value of close collaboration between research labs and accredited diagnostic laboratories, like CENTOGENE, in finding precise molecular diagnosis for families affected by undiagnosed ultra-rare disorders. These collaborations are fundamental in advancing our understanding of genetic disorders and their underlying mechanisms."

Prof. Peter Bauer, Chief Genomic Officer at CENTOGENE, added, "Up until now, the genetic causes of neurological disorders have remained greatly unknown. This groundbreaking study has now not only helped us to understand a driving factor for 45 patients but has shed light on the way to a potential cure for a larger group of neurological disorders, and that is game changing. That is what we are striving for every day."

To read the full study, visit: <https://link.centogene.com/Brain-Publication>

About CENTOGENE

CENTOGENE's mission is to provide data-driven, life-changing answers to patients, physicians, and pharma companies for rare and neurodegenerative diseases. We integrate multiomic technologies with the CENTOGENE Biodatabank – providing dimensional analysis to guide the next generation of precision medicine. Our unique approach enables rapid and reliable diagnosis for patients, supports a more precise physician understanding of disease states, and accelerates and de-risks targeted pharma drug discovery, development, and commercialization.

Since our founding in 2006, CENTOGENE has been offering rapid and reliable diagnosis – building a network of approximately 30,000 active physicians. Our ISO, CAP, and CLIA certified multiomic reference laboratories in Germany utilize Phenomic, Genomic, Transcriptomic, Epigenomic, Proteomic, and Metabolomic datasets. This data is captured in our CENTOGENE Biodatabank, with over 800,000 patients represented from over 120 highly diverse countries, over 70% of whom are of non-European descent. To date, the CENTOGENE Biodatabank has contributed to generating novel insights for more than 285 peer-reviewed publications.

By translating our data and expertise into tangible insights, we have supported over 50 collaborations with pharma partners. Together, we accelerate and de-risk drug discovery, development, and commercialization in target and drug screening, clinical development, market access and expansion, as well as offering CENTOGENE Biodata Licenses and Insight Reports to enable a world healed of all rare and neurodegenerative diseases.

To discover more about our products, pipeline, and patient-driven purpose, visit www.centogene.com and follow us on [LinkedIn](#).

Forward-Looking Statements

This press release contains “forward-looking statements” within the meaning of the U.S. federal securities laws. Statements contained herein that are not clearly historical in nature are forward-looking, and the words “anticipate,” “believe,” “continue,” “expect,” “estimate,” “intend,” “project,” “plan,” “is designed to,” “potential,” “predict,” “objective” and similar expressions and future or conditional verbs such as “will,” “would,” “should,” “could,” “might,” “can,” and “may,” or the negative of these are generally intended to identify forward-looking statements. Such forward-looking statements involve known and unknown risks, uncertainties, and other important factors that may cause CENTOGENE’s actual results, performance, or achievements to be materially different from any future results, performance, or achievements expressed or implied by the forward-looking statements. Such risks and uncertainties include, among others, negative economic and geopolitical conditions and instability and volatility in the worldwide financial markets, possible changes in current and proposed legislation, regulations and governmental policies, pressures from increasing competition and consolidation in our industry, the expense and uncertainty of regulatory approval, including from the U.S. Food and Drug Administration, our reliance on third parties and collaboration partners, including our ability to manage growth, execute our business strategy and enter into new client relationships, our dependency on the rare disease industry, our ability to manage international expansion, our reliance on key personnel, our reliance on intellectual property protection, fluctuations of our operating results due to the effect of exchange rates, our ability to streamline cash usage, our continued ongoing compliance with covenants linked to financial instruments, our requirement for additional financing, and our ability to continue as a going concern, or other factors. For further information on the risks and uncertainties that could cause actual results to differ from those expressed in these forward-looking statements, as well as risks relating to CENTOGENE’s business in general, see CENTOGENE’s risk factors set forth in CENTOGENE’s Form 20-F filed on May 16, 2023, with the Securities and Exchange Commission (the “SEC”) and subsequent filings with the SEC. Any forward-looking statements contained in this press release speak only as of the date hereof, and CENTOGENE specifically disclaims any obligation to update any forward-looking statement, whether as a result of new information, future events, or otherwise.

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