CENTOGENE Discovers Six New Rare Diseases by Leveraging the Strength of Its Bio/Databank

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Research leads to diagnosis and treatment options for rare disease patients

- More than half of patients with genetic diseases remain undiagnosed, even after performing Exome and Genome Sequencing
- By performing deep genetic analyses and Bio/Databank mining, CENTOGENE discovers six novel gene-disease associations and evidence supporting 31 candidate genes
- Study reveals value of such Bio/Databanks in diagnosing and accelerating treatment options for rare disease patients

CAMBRIDGE, Mass. and ROSTOCK, Germany and BERLIN, May 05, 2021 (GLOBE NEWSWIRE) -- Centogene N.V. (Nasdaq: CNTG), a commercial-stage company focused on rare diseases that transforms real-world clinical and genetic data into actionable information for patients, physicians, and pharmaceutical companies, announced today research revealing six novel gene-disease associations for a wide range of genetic disorders and confirmation of 31 additional candidate genes. The findings are a result of in-depth analyses into the Company's rare disease Bio/Databank, after standard genetic testing was unable to determine the exact cause of symptoms. As a result, over 90 patients were able to finally receive a diagnosis – opening the potential to diagnose countless others following further research. Additionally, these findings have revealed potential treatment options for patients based on known-disease overlaps.

The landmark study's findings have been published in *Genetics in Medicine* (https://www.nature.com/articles/s41436-021-01159-0) – the official Journal of the American College of Medical Genetics and Genomics.

Prof. Peter Bauer, Chief Genomic Officer at CENTOGENE, said, "There are more than 7,000 rare diseases that are known to date, and our research shows that there are even more to be identified. We see it as our responsibility to dive deeper into patient information when carrying out genetic testing. In this case, we were able to provide patients with a diagnosis with diseases that were not previously on anyone's radar. Even more, we were able to help improve their quality of life – opening up treatment and management options."

"Although technological advancements over the past decade have led to improved diagnostics, our study shows that simply applying a genetic test is not enough," adds Dr. Aida Bertoli-Avella, Head of Research Data Analysis. "Especially when it comes to rare diseases, you have to look past the surface – using advanced tools to reveal the underlying cause of a disease and open up a new world of treatment options."

The research led to over 90 patients receiving a diagnosis based on the six new revealed rare diseases and 31 candidate genes – gaining access to future treatment options and improved medical management.

About the Study

While technology has advanced over the past ten years, more than half of patients with genetic diseases remain undiagnosed, even after applying genome-wide diagnostic approaches. The challenge of variant interpretation remains at the forefront of diagnostic challenges, in part due to the missing gene-phenotype link.

Leveraging CENTOGENE's rare disease Bio/Databank, the Company carried out an extensive Exome Sequencing and Genome Sequencing evaluation of genes with no known disease association and patients suffering genetic diseases that had remained undiagnosed. From there, the Company performed analyses on its biobank using specific criteria, which enabled further identification of unrelated patients displaying similar phenotypes. Ultimately, this led to the discovery of six novel gene-disease associations based on 38 severely affected patients with variants in six genes: *BLOC1S1*, *IPO8*, *MMP15*, *PLK1*, *RAP1GDS1*, and *ZNF699,* as well as the elucidation of 31 additional candidate genes.

To read the complete study in *Genetics in Medicine*, visit: https://www.nature.com/articles/s41436-021- 01159-0

About CENTOGENE

CENTOGENE engages in diagnosis and research around rare diseases transforming real-world clinical and genetic data into actionable information for patients, physicians, and pharmaceutical companies. Our goal is to bring rationality to treatment decisions and to accelerate the development of new orphan drugs by using our extensive rare disease knowledge, including epidemiological and clinical data, as well as innovative biomarkers. CENTOGENE has developed a global proprietary rare disease platform based on our real-world data repository with over 3.9 billion weighted data points from approximately 600,000 patients representing over 120 different countries as of December 31, 2020.

The Company's platform includes epidemiologic, phenotypic, and genetic data that reflects a global population, and also a biobank of these patients' blood samples. CENTOGENE believes this represents the only platform that comprehensively analyzes multi-level data to improve the understanding of rare hereditary diseases, which can aid in the identification of patients and improve our pharmaceutical partners' ability to bring orphan drugs to the market. As of December 31, 2020, the Company collaborated with over 30 pharmaceutical partners.

Important Notice and Disclaimer

This press release contains statements that constitute “forward-looking statements” as that term is defined in the United States Private Securities Litigation Reform Act of 1995, including statements that express the Company’s opinions, expectations, beliefs, plans, objectives, assumptions, or projections regarding future events or future results, in contrast with statements that reflect historical facts. Examples include discussion of our strategies, financing plans, growth opportunities, and market growth. In some cases, you can identify such forward-looking statements by terminology such as “anticipate,” “intend,” “believe,” “estimate,” “plan,” “seek,” “project” or “expect,” “may,” “will,” “would,” “could,” or “should,” the negative of these terms or similar expressions. Forward-looking statements are based on management’s current beliefs and assumptions and on information currently available to the Company. However, these forward-looking statements are not a guarantee of our performance, and you should not place undue reliance on such statements. Forward-looking statements are subject to many risks, uncertainties, and other variable circumstances, such as negative worldwide economic conditions and ongoing instability and volatility in the worldwide financial markets, the effects of the COVID-19 pandemic on our business and results of operations, 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 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, or other factors. Such risks and uncertainties may cause the statements to be inaccurate and readers are cautioned not to place undue reliance on such statements. Many of these risks are outside of the Company’s control and could cause its actual results to differ materially from those it thought would occur. The forward-looking statements included in this press release are made only as of the date hereof. The Company does not undertake, and specifically declines, any obligation to update any such statements or to publicly announce the results of any revisions to any such statements to reflect future events or developments, except as required by law.

For further information, please refer to the Risk Factors section in our Annual Report for the year ended December 31, 2020, on Form 20-F filed with the SEC on April 15, 2021, and other current reports and documents furnished to or filed with the U.S. Securities and Exchange Commission (SEC). You may get these documents by visiting EDGAR on the SEC website at www.sec.gov.

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