



CENTOGENE Signs New Collaboration with PTC Therapeutics for Global Diagnostic Testing Program

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Focus on Aromatic L-amino Acid Decarboxylase (AADC) Deficiency

Cambridge, MA, USA & Rostock, Berlin, GERMANY, 18 November 2019 – 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 that it has signed an agreement with PTC Therapeutics, Inc. (PTC) for a global diagnostic program for aromatic L-amino acid decarboxylase (AADC) deficiency.

The testing program for AADC deficiency will provide physicians with much needed analysis of 3-O-Methyldopa (3OMD), a powerful metabolic biomarker measured by mass spectrometry. When indicated by abnormal 3OMD levels, next generation sequencing (NGS) of the dopa decarboxylase (DDC) gene and variant analysis – providing 100% coverage – will be conducted. Eventually, deletion/duplication analysis will then be run if no mutation is identified via NGS.

Analysis will be performed using CentoCard® – CENTOGENE's CE-marked dried blood spot collection kit – that will be directly shipped to physicians. CENTOGENE also is in the process of validating DDC enzyme activity from dried blood spot and in doing so aims to offer 3OMD, NGS and DDC enzyme activity testing using a single CentoCard®. This represents a complete set of assays facilitating the diagnosis of AADC.

"We are pleased to support PTC Therapeutics for this important diagnostic program helping patients suffering from symptoms related to AADC deficiency. We aim to provide physicians with a consensus diagnosis that includes a biomarker, and genetic validation from a single source," said Dr. Arndt Rolfs, CEO CENTOGENE. "The analysis of 3-O-Methyldopa (3OMD) based on our dried blood spot test at CENTOGENE underscores our passion for helping to reduce the diagnostic odyssey and for bringing hope to patients and their families."

AADC deficiency is a rare inherited disorder that affects the way signals are passed between certain cells in the nervous system. It causes severe developmental disabilities, the inability to develop any motor strength and control, frequent hospitalizations, and the need for life-long care.

Questions regarding diagnostic testing for AADC deficiency can be directed to [AADCTesting\(at\)ptcbio\(dot\)com](mailto:AADCTesting(at)ptcbio(dot)com) For more information on AADC deficiency, please visit the following websites:

- Healthcare professionals – <https://aadcsinsights.com>
- Caregivers or patients – <https://aboutaadc.com>