PTC Therapeutics Launches A No-Cost Testing Program to Drive AADC Deficiency Patient Identification

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- AADC deficiency patients are often misdiagnosed as having other neurological conditions such as cerebral palsy -

SOUTH PLAINFIELD, N.J., Nov. 18, 2019 /PRNewswire/ -- PTC Therapeutics, Inc. (NASDAQ: PTCT) today announced the launch of a no-cost testing program for aromatic L-amino acid decarboxylase (AADC) deficiency, a rare genetic condition. Children with AADC deficiency fail to thrive and commonly don't reach developmental milestones that include being able to hold up their head, sit unassisted, and stand.[1] These patients also have muscular hypotonia, severe seizure-like episodes that include oculogyric crises, and the need for intensive life-long care.[1] To raise awareness and identify misdiagnosed AADC deficiency patients, PTC has launched a no-cost testing program to genetically distinguish these patients from those with other disorders.

"We know that there are a large number of patients with AADC deficiency that have yet to be properly diagnosed," said Stuart W. Peltz, Ph.D., Chief Executive Officer of PTC Therapeutics. "For many AADC deficiency patients and their families, the journey to diagnosis is extremely challenging. Many patients spend years without a diagnosis or are misdiagnosed with other neurological conditions, such as cerebral palsy. We are very proud to provide diagnostic testing – at no cost to the AADC deficiency community – to support accurate and earlier diagnosis for patients."

AADC deficiency is caused by pathogenic mutations in the dopa decarboxylase (*DDC*) gene, resulting in a lack of functioning AADC enzyme, which is responsible for the final step in the synthesis of key neurotransmitters dopamine and serotonin.[1] In the brain, dopamine and serotonin are important nerve cell signalling molecules.

"Identifying the underlying genetic mutations of AADC deficiency is critical to understand the nature and prognosis of an individual patient's disease, and to ensure the best possible treatment," said Dr. Arndt Rolfs, CEO CENTOGENE. "The analysis of 3-O-Methyldopa (3OMD) based on an innovative dried blood spot test at CENTOGENE is extremely important in support of patients suffering from symptoms related to AADC deficiency, and underscores our passion for helping to reduce the diagnostic odyssey and bring hope to patients and their families."

The free test requires only a blood sample from a patient, which is analyzed to identify a number of indicators for the disabling disease. Healthcare professionals can order the test from PTC if they have concerns about patients who may be showing symptoms of AADC deficiency.

Questions regarding diagnostic testing for AADC deficiency can be directed to AADCDtesting@ptcbio.com. For more information on AADC deficiency, please visit the websites below:

- Healthcare professionals – https://aadcinsights.com/
- Caregivers or patients – https://aboutaadc.com/

About aromatic L-amino acid decarboxylase (AADC) deficiency
Aromatic L-amino acid decarboxylase (AADC) deficiency is a rare genetic condition caused by pathogenic mutations in the dopa decarboxylase (*DDC*) gene, resulting in a lack of functioning AADC enzyme, which is responsible for the final step in the synthesis of key neurotransmitters dopamine and serotonin.[1]

AADC deficiency results in delays or failure to reach developmental milestones such as walking, talking, sitting unassisted, or having head control, low muscle tone (also known as muscular hypotonia), severe, seizure-like episodes involving involuntary eye rolling (also known as oculogyric crises), autonomic abnormalities, and the need for life-long care.[1] Given this neurologically devastating illness, patients with severe AADC deficiency have a high risk for death during childhood. Treatment options for patients with AADC deficiency are limited and there are currently no approved therapies that address the underlying cause.

About the AADC deficiency no-cost test
Initial testing for AADC deficiency has previously, typically involved an analysis of cerebrospinal fluid (CSF), taken via a lumbar puncture, also known as a spinal tap. No-cost testing supported by PTC Therapeutics involves a simple blood test to screen for increased levels of 3-OMD, a neurotransmitter metabolite that is elevated in patients with AADC deficiency. If 3-OMD levels are elevated, then an AADC enzyme activity analysis and DDC gene sequencing are conducted to confirm the diagnosis.

Outside of the U.S., if 3-OMD levels are elevated, DDC gene sequencing is conducted to confirm diagnosis. Country-specific rules regarding the test may differ, and restrictions may apply based on the country from where the request originates.

About PTC Therapeutics, Inc.
PTC is a science-driven, global biopharmaceutical company focused on the discovery, development and commercialization of clinically-differentiated medicines that provide benefits to patients with rare disorders. PTC's ability to globally commercialize products is the foundation that drives investment in a robust pipeline of transformative medicines and our mission to provide access to best-in-class treatments for patients who have an unmet medical need. To learn more about PTC, please visit us on www.ptcbio.com and follow us on Facebook, on Twitter at @PTCBio, and on LinkedIn.

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2. Hwu et al. Natural History of Aromatic L-Amino Acid Decarboxylase Deficiency in Taiwan. JIMD Rep. 2018; 40: 1-6.
3. Wassenberg et al. Consensus guideline for the diagnosis and treatment of aromatic l-amino acid decarboxylase (AADC) deficiency. Orphanet J Rare Dis. 2017; 12:12.

Forward-Looking Statements:
This press release contains forward-looking statements within the meaning of The Private Securities Litigation Reform Act of 1995. All statements contained in this release, other than statements of historic fact, are forward-looking statements, including statements regarding: the future expectations, plans and prospects for PTC; expectations with respect to PTC's gene therapy platform, including any potential regulatory submissions; PTC's strategy, future operations, future financial position, future revenues, projected costs; and the objectives of management. Other forward-looking statements may be identified by the words "guidance", "plan," "anticipate," "believe," "estimate," "expect," "intend," "may," "target," "potential," "will," "would," "could," "should," "continue," and similar expressions.

PTC's actual results, performance or achievements could differ materially from those expressed or implied by forward-looking statements it makes as a result of a variety of risks and uncertainties, including those related to: expectations with respect to its gene therapy platform, including expectations with respect to the potential achievement of development, regulatory and sales milestones and contingent payments to the former Agilis Biotherapeutics, Inc. equityholders with respect thereto and PTC's ability to obtain marketing approval of PTC-AADC and other gene therapy product candidates will not be realized or will not be realized within the expected time period; significant transaction costs, unknown liabilities, the risk of litigation and/or regulatory actions related to the acquisition of its gene therapy pipeline, as well as other business effects, including the effects of industry, market, economic, political or regulatory conditions; the eligible patient base and commercial potential of PTC-AADC; and the factors discussed in the "Risk Factors" section of PTC's most recent Annual Report on Form 10-K, as well as any updates to these risk factors filed from time to time in PTC's other filings with the SEC. You are urged to carefully consider all such factors.

As with any pharmaceutical under development, there are significant risks in the development, regulatory approval and commercialization of new products. There are no guarantees that any product will receive or maintain regulatory approval in any territory, or prove to be commercially successful, including PTC-AADC.

The forward-looking statements contained herein represent PTC's views only as of the date of this press release and PTC does not undertake or plan to update or revise any such forward-looking statements to reflect actual results or changes in plans, prospects, assumptions, estimates or projections, or other circumstances occurring after the date of this press release except as required by law.

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