PTC Therapeutics Launches PTC PINPOINT - A Genetic Testing Program for Individuals Suspected of Having Neurotransmitter Disorders

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- Early diagnosis through genetic testing may lead to improved long-term outcomes for patients with neurotransmitter disorders, including AADC deficiency -

SOUTH PLAINFIELD, N.J., Sept. 9, 2020 /PRNewswire/ -- PTC Therapeutics, Inc. (NASDAQ: PTCT) today introduced a new program that provides genetic testing and counseling for people who may have a neurotransmitter disorder, such as aromatic L-amino acid decarboxylase (AADC) deficiency. The program, called PTC PINPOINT, will offer the testing at no charge to patients which can lead to an earlier diagnosis and treatment.

"Genetic testing programs in rare disorders are crucial for more rapid and accurate diagnosis so that patients can be identified early and receive treatment as young as possible," said Stuart W. Peltz, Ph.D., Chief Executive Officer of PTC Therapeutics. "We have joined forces with Invitae to eliminate financial barriers to genetic testing for patients with neurotransmitter disorders. The PINPOINT program includes genetic testing for patients suffering from AADC deficiency, a genetic disorder for which PTC is developing an innovative gene therapy."

PTC PINPOINT is being conducted in partnership with Invitae, (NYSE: NVTA), and will be available at no charge to patients in the United States. who are suspected of having symptoms consistent with a neurotransmitter disorder, such as tyrosine hydroxylase (TH) deficiency, AADC deficiency, dopamine β-hydroxylase deficiency and others that are considered genetically heterogeneous, affect the metabolism, and movement of neurotransmitters.[1] These disorders are typically present in infancy or childhood, and can include development problems, hypotonia, early onset parkinsonism, dystonia, ataxia, autonomic dysfunction, oculogyric crises, epilepsy and more. In certain cases, treatment and clinical trials are available for some of these disorders, and early diagnosis may help improve long-term outcomes.

PTC PINPOINT uses the Invitae Neurotransmitter Disorders Panel, which analyzes up to 37 genes that are associated with disorders of monoamine metabolism, GABA metabolism, and neurotransmitter receptors and transporters.

Healthcare professionals can order the test from PTC PINPOINT if their patients are showing symptoms of neurotransmitter diseases. Additional details, terms and conditions of the program can be found at www.invitae.com/ptc-pinpoint.

Individuals tested through PTC PINPOINT are eligible for post-test genetic counselling to help them understand their test results. This service is provided through GeneMatters, a third-party genetic counselling service, and is made available by Invitae at no charge.

"Genetic testing can expedite making an accurate diagnosis, facilitate earlier interventions, allow genetic counseling of family members, and support clinical research into neurotransmitter disorders. Early testing is essential, as many neurodegenerative diseases have similar clinical presentations but will likely differ in which treatment is appropriate once they become available. Furthermore, there is ample experience with other rare neurodegenerative disorders that outcomes will most likely be better the sooner treatment is started," said Robert Nussbaum, M.D., Chief Medical Officer of Invitae. "We believe this partnership with PTC Therapeutics will help increase access to early genetic testing to provide answers and inform care for many patients for whom early intervention can make all the difference."

About aromatic L-amino acid decarboxylase (AADC) deficiency

Aromatic L-amino acid decarboxylase (AADC) deficiency is a rare genetic condition caused by a mutation 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.[2]

AADC deficiency results in delays or failure to reach developmental milestones such as head control, sitting, standing, walking, or talking, low muscle tone (also known as muscular hypotonia), severe, seizure-like episodes involving involuntary eye movement (also known as oculogyric crises), autonomic abnormalities, and the need for life-long care.[2] Given this neurologically devastating illness, patients with severe AADC deficiency have a high risk of death during childhood. There are currently no approved therapies that address the underlying cause.

About Invitae

Invitae Corporation (NYSE: NVTA) is a leading medical genetics company, whose mission is to bring comprehensive genetic information into mainstream medicine to improve healthcare for billions of people. Invitae's goal is to aggregate the world's genetic tests into a single service with higher quality, faster turnaround time, and lower prices. For more information, visit the company's website at invitae.com.

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 and diversified 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 at www.ptcbio.com and follow us on Facebook, on Twitter at @PTCBio, and on LinkedIn.

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References:

1. Jaeken et al. Disorders of Neurotransmission. Inborn Metabolic Diseases. 2006.
2. 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; PTC's strategy, future operations, future financial position, future revenues and 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: the eligible patient base and commercial potential of PTC's products and product candidates; the outcome of pricing, coverage and reimbursement negotiations with third party payors for PTC's products or product candidates that PTC commercializes or may commercialize in the future; significant business effects, including the effects of industry, market, economic, political or regulatory conditions; changes in tax and other laws, regulations, rates and policies; PTC's scientific approach and general development progress; and the factors discussed in the "Risk Factors" section of PTC's most recent Quarterly Report on Form 10-Q and 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.

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.