



PTC Therapeutics Launches the PTC Pinpoint Direct™ Program for Patient-Initiated Genetic Testing

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- Provides easier access to genetic testing for patients with symptoms or diagnosis of cerebral palsy at no charge -

SOUTH PLAINFIELD, N.J., April 14, 2021 /PRNewswire/ -- PTC Therapeutics, Inc. (NASDAQ: PTCT) today introduced a new no-charge, patient-initiated testing program in the United States for individuals who have symptoms consistent with, or a diagnosis of, cerebral palsy (CP) with no evidence of brain injury.

CP is a group of disorders that affect a person's ability to move and maintain balance and posture. Many of the common symptoms of CP are similar to other neurotransmitter disorders, such as Aromatic L-amino Acid Decarboxylase (AADC) deficiency, resulting in potential misdiagnosis. The program, PTC Pinpoint Direct™-CP Spectrum, is an innovative, genetic testing program which includes expert genetic counseling at no charge for eligible patients.

"An umbrella diagnosis of a neurotransmitter disorder, such as cerebral palsy, can be extremely frustrating, especially for patients and caregivers who spend years with unexplained and underlying symptoms," said Kylie O'Keefe, Senior Vice President, Global Commercial and Corporate Strategy, PTC Therapeutics. "Up to one-third of cerebral palsy cases are driven by a genetic disorder, and some of those conditions may have transformative treatments currently available. We are excited to launch this new extension of our existing program, PTC PINPOINT, to further eliminate the barriers of genetic testing for rare diseases."

PTC Pinpoint Direct™-CP Spectrum will be available at no charge to patients of all ages in the United States who have symptoms consistent with, or a diagnosis of, CP without evidence of brain injury. Testing with PTC Pinpoint Direct™-CP Spectrum may help uncover a genetic cause of the symptoms and determine the right plan for the patient's care, including new and emerging treatment options. PTC Pinpoint Direct™-CP Spectrum may also eliminate unnecessary treatments or medications that may cause complications.

The program is sponsored by PTC Therapeutics and is the first collaboration between PTC, Genome Medical for expert genetic counseling and administration, and Invitae for industry-leading genetic testing. Caregivers and patients may initiate the testing process from PTC Pinpoint Direct online and receive information on eligibility requirements. Additional details can be found at www.PTCPinpointDirect.com.

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 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 Genome Medical

Genome Medical is a digital health company that is transforming the delivery of genetic and genomics care through telehealth and digital infrastructure innovations. It operates as an independent virtual medical practice, with on-demand genetic expertise, testing recommendations and patient navigation platforms that accelerate the delivery of genomic insights as a service. To learn more, visit genomemedical.com and follow @GenomeMed.

About PTC

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. The Company's strategy is to leverage its strong scientific expertise and global commercial infrastructure to maximize value for its patients and other stakeholders. To learn more about PTC, please visit us at www.ptcbio.com and follow us on Facebook, Instagram, LinkedIn and on Twitter at @PTCBio.

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.

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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, 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 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.

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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