Results Show Long-Lasting and Holistic Improvements in Children with AADC Deficiency Treated with PTC-AADC Gene Therapy

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- Analysis of Five-Year Results Presented at 50th Child Neurology Society Annual Meeting -

SOUTH PLAINFIELD, N.J., Sept. 29, 2021 /PRNewswire/ -- PTC Therapeutics, Inc. (NASDAQ: PTCT) today presented a new analysis of five-year results that shows its novel gene therapy, PTC-AADC, leads to profound improvements in children with aromatic L-Amino acid decarboxylase (AADC) deficiency, a previously intractable, fatal and devastating rare disorder of the central nervous system.[1-4] Children treated with PTC-AADC developed motor function and cognitive skills not previously seen, such as holding up their head, sitting or standing with support and communicating, and these persisted for up to 10 years.[1]

"The transformations we are seeing in the children are remarkable, as they could not even lift their heads before treatment," said Stuart W. Peltz, Ph.D., Chief Executive Officer, PTC Therapeutics. "Treatment with PTC-AADC is restoring dopamine production, which leads to the children now being able to sit, roll over, stand and walk. They are also able to communicate, and gain both weight and strength. We believe these results show its potential as a transformational therapy for the AADC patients and their families."

The new, five-year results show that motor function improvements were sustained, demonstrating that the treatment effect of PTC-AADC is durable, which is highly important for a single administration therapy for a chronic, fatal disease.[1-2] Across three clinical trials, improvements in motor development were recorded in all children from as early as 3 months.[1] Cognitive and language skills were also reported to improve significantly from baseline, as measured by Bayley-III scores, with children able to understand their caregivers and express themselves.[2]

In addition, the rate of respiratory infection declined from an average of 2.4 episodes per year at 12 months to 0.6 episodes per year at 2 years and 0.3 episodes per year at 5 years.[3] Almost all treated children went from a baseline weight below the third percentile to making age-appropriate weight gains by 12 months following gene therapy treatment.[3]

There are currently no approved disease-modifying therapies for AADC deficiency, a fatal disorder caused by a defective gene that stops the body from producing the dopamine needed to develop and function normally. Delivering PTC-AADC gene therapy directly to the putamen, a site in the brain where dopamine is utilized, corrects this defect.[4]

"The impact that this one-time gene therapy has had on children with AADC deficiency in these trials is truly life-changing," said investigator Paul Wuh-Liang Hwu, National Taiwan University Hospital. "We are encouraged by the strong safety profile and long-lasting effect seen with PTC-AADC, and how it has improved the lives of patients and their caregivers."

PTC-AADC is currently under review by the European Medicines Agency's Committee for Medicinal Products for Human Use with an opinion expected in the fourth quarter of 2021, and is an investigational new drug in the United States. A Biologics License Application is expected to be submitted to the U.S. Food and Drug Administration by the end of 2021.

About aromatic L-amino acid decarboxylase (AADC) deficiency
AADC deficiency is a fatal, rare genetic disorder that causes severe disability and suffering from the first months of life, affecting every aspect of life – physical, mental, and behavioral. The suffering of children with AADC deficiency is exacerbated by episodes of distressing seizure-like oculogyric crises, which can happen daily and last for hours, causing the eyes to roll up in the head, frequent vomiting, behavioral problems, difficulty sleeping, and life-threatening complications such as respiratory infections and gastrointestinal problems.

Current management options yield limited improvement for the majority of patients with AADC deficiency. Managing patients with AADC deficiency

requires a multidisciplinary team of specialists and complex coordination of care to address significant health issues, including developmental delays, infections, orthopaedic and cardiac complications, and other comorbidities

While several diagnostic tests for AADC deficiency are available, the condition remains largely undiagnosed.

About PTC Therapeutics, Inc.
PTC Therapeutics 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 mission is to provide access to best-in-class treatments for patients with an unmet medical need, using its ability to globally commercialize products as the foundation to drive investment in a robust and diversified pipeline of transformative medicines. 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 it on Facebook, on Twitter at @PTCBio, and on LinkedIn.

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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, including with respect to the expected timing of clinical trials and studies, availability of data, regulatory submissions and responses and other matters; expectations with respect to PTC's gene therapy platform, including any regulatory submissions and manufacturing capabilities; PTC's expectations with respect to the licensing, regulatory submissions and commercialization of its other products and product candidates; 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: 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; expectations with respect to PTC's gene therapy platform, including any regulatory submissions and potential approvals, manufacturing capabilities and the potential financial impact and benefits of its leased biologics manufacturing facility and the potential achievement of development, regulatory and sales milestones and contingent payments that PTC may be obligated to make; significant business effects, including the effects of industry, market, economic, political or regulatory conditions; changes in tax and other laws, regulations, rates and policies; the eligible patient base and commercial potential of PTC's products and product candidates; PTC's scientific approach and general development progress; 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.

[1] Hwu PW-L, *et al.* Eladocagene Exuparvovec Gene Therapy Improves Motor Development in Patients With Aromatic L-Amino Acid Decarboxylase Deficiency. Poster presented at the CNS Annual Meeting; October 16-20, 2021.

[2] Hwu PW-L, *et al.* Gene Therapy With Eladocagene Exuparvovec Improves Cognition and Language in Patients With Aromatic L-Amino Acid Decarboxylase Deficiency. Poster presented at the CNS Annual Meeting; October 16-20, 2021.

[3] Hwu PW-L, *et al.* Eladocagene Exuparvovec Improves Body Weight and Reduces Respiratory Infections in Patients With Aromatic L-Amino Acid Decarboxylase Deficiency. Poster presented at the CNS Annual Meeting; October 16-20, 2021.

[4] Chien YH, *et al.* Efficacy and safety of AAV2 gene therapy in children with aromatic L-amino acid decarboxylase deficiency: an open-label, phase 1/2 trial. *Lancet Child Adolesc Health.* 2017;1(4):265-273.

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