



New Data Shows Devastating Impact of AADC Deficiency on Caregivers

May 17, 2021

- Caregivers spend on average 15 hours a day providing care -

SOUTH PLAINFIELD, N.J., May 17, 2021 /PRNewswire/ -- PTC Therapeutics, Inc. (NASDAQ: PTCT) today announced data from the first study on the impact of caring for a child with the ultra-rare genetic disorder, aromatic L-Amino acid decarboxylase deficiency (AADC-d). Results show that caregivers spend almost every waking moment caring for a child with AADC-d, requiring most to leave their jobs or reduce their working hours.¹ The data presented at the Virtual ISPOR North America 2021 Conference detail new insights into the extent of the burden of AADC-d and the great need and value of effective treatments.

"As with so many rare diseases, there is a tremendous impact endured by a family caring for a child with AADC deficiency," said Kylie O'Keefe, Senior Vice President, Global Commercial and Corporate Strategy, PTC Therapeutics, Inc. "These insights demonstrate the high unmet need for therapies and the associated benefits a treatment could provide to caregivers and their families."

AADC-d is a fatal, ultra-rare, genetic disorder that causes severe disability and suffering, and the need for 24-hour care.²⁻⁵ The study found that primary caregivers spent on average 15 hours a day (mean of 105 hours per week) on practical and emotional care and carrying out administrative tasks like planning and attending appointments.¹

The study showed that 75% of caregivers left their jobs or greatly reduced their working hours, and 55% needed additional help (paid or unpaid) to supplement the care they were providing to their children.¹ Unpaid support usually came from a partner who provided, on average, 37 hours of help a week (8-93 hours per week).¹ Families who required paid support received, on average, 27 additional hours of help (10-35 hours per week) from a registered nurse or training nursing assistant.¹

Data from a PTC health utility study was also presented orally. The study, which was based on an online survey of 1,000 respondents from the French general public, showed that the utility values increased as the health of a child with AADC deficiency improved.⁶ These data highlight the value of treating AADC-d and the pain and distress of its symptoms.⁶ This work builds on PTC's previous AADC health utility data presented at [ISPOR Europe 2020](#).

For More Information:

Investors

Kylie O'Keefe
+1 (908) 300-0691
kokeefe@ptcbio.com

Media

Jane Baj
+1 (908) 912-9167
jbaj@ptcbio.com

About aromatic L-amino acid decarboxylase (AADC) deficiency

AADC deficiency is a fatal, ultra-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, behavioural problems, difficulty sleeping, and life-threatening complications such as respiratory infections and gastrointestinal problems.

There are no effective treatments for AADC deficiency, and the lives of affected children are highly medicalized, sometimes involving many different medications to help manage symptoms, ongoing physical, occupational and speech therapy, and interventions, including surgery, to manage potentially life-threatening complications (infections, severe feeding and breathing problems and scoliosis).

While several diagnostic tests for AADC deficiency are available, the condition remains largely undiagnosed or misdiagnosed for other conditions with similar symptoms, such as cerebral palsy and some forms of epilepsy.

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](#).

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 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, 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.

References:

1. Buesch K, Williams K, *et al.* Caring for an Individual with Aromatic L-amino Acid Decarboxylase (AADC) Deficiency: Analysis of Reported Time for Practical and Emotional Care and Paid/Unpaid Help. *Poster presented at Virtual ISPOR North America 2021 Conference.*
2. Wassenberg T, *et al.* Consensus guideline for the diagnosis and treatment of aromatic l-amino acid decarboxylase (AADC) deficiency. *Orphanet J Rare Dis.* 2017;12(1):12.
3. Hwu WL *et al.* Natural History of Aromatic L-Amino Acid Decarboxylase Deficiency in Taiwan. *JIMD Rep.* 2018; 40:1-6.
4. Himmelreich N, *et al.* Aromatic amino acid decarboxylase deficiency: Molecular and metabolic basis and therapeutic outlook. *Mol Genet Metab.* 2019;127(1):12-22.
5. Chien YH, *et al.* 3-O-methyldopa levels in newborns: Result of newborn screening for aromatic l-amino-acid decarboxylase deficiency. *Mol Genet Metab.* August 2016;118(4):259-263.
6. Smith AB, Hanbury A, *et al.* A Discrete Choice Experiment to Derive Health State Utilities for Aromatic L-amino Acid Decarboxylase (AADC) Deficiency in France. *Poster presented at Virtual ISPOR North America 2021 Conference*

 View original content: <http://www.prnewswire.com/news-releases/new-data-shows-devastating-impact-of-aadc-deficiency-on-caregivers-301292447.html>

SOURCE PTC Therapeutics, Inc.