



PTC Therapeutics Highlights Data on High Disease Burden and Value of Treatment Across Two Rare Diseases

November 16, 2020

- AADC deficiency associated with high and accumulating disease burden, demonstrating the need for access to an effective therapy -

- Patient and caregiver quality of life study reveals real-life impact of nonsense mutation Duchenne muscular dystrophy and reinforces value of treatment -

SOUTH PLAINFIELD, N.J., Nov. 16, 2020 /PRNewswire/ -- PTC Therapeutics, Inc. (NASDAQ: PTCT) today announced the presentation of data on two rare diseases, providing new insights on the burden of Aromatic L-Amino acid Decarboxylase deficiency (AADC-d) and Duchenne muscular dystrophy caused by nonsense mutation (nmDMD). The data, presented at the Virtual ISPOR Europe 2020 Conference, further advance the understanding of these rare and devastating childhood genetic disorders, and the need and value of effective treatments.

"PTC has been working with the rare disease community to better understand the impact of rare diseases on patients and their families for over 20 years," said Stuart W. Peltz, Ph.D., Chief Executive Officer, PTC Therapeutics. "These insights are critical so that the best treatments can be made available to patients. PTC is committed to ensuring patients can access clinically differentiated treatments such as our gene therapy for AADC deficiency which, if approved, promises to become the standard of care."

AADC-d associated with high and accumulating disease burden, demonstrating the need for access to an effective therapy

AADC-d is an ultra-rare, genetic disorder that causes severe disability, and the need for 24-hour care and high mortality.¹ There is currently no effective disease-modifying therapy approved for patients with AADC-d. PTC-AADC (eladocogene exuparvovec) is the first treatment for AADC-d under regulatory review. The Marketing Authorization Application was submitted to the European Medicines Agency (EMA) in January 2020, and the Committee for Medicinal Products for Human Use (CHMP) opinion is expected in the first half of 2021.

A review of 66 patient case reports found that symptoms of AADC-d that contribute to high disease burden started from birth (in 29 percent of cases) and accumulated over time.² By age 6, 28 percent of patients were so severely disabled that they experienced symptoms in six core domains.*

Further data from 28 patients who had received PTC's investigational gene therapy, PTC-AADC was presented, including data from three clinical trials with follow-up from one to nine years, predicting the long-term and durable benefits of the therapy,³ including future motor milestone achievement, by using the Peabody Developmental Motor Scales assessment (PDMS-2) score.

New insights into the impact of nmDMD and treatment from patient and carer perspectives^{4,5,6}

nmDMD is a rare and fatal genetic disorder that results in progressive muscle weakness from early childhood, and leads to premature death in the mid-twenties due to heart and respiratory failure.⁷ PTC provides access to two of the first drugs approved for nmDMD in the world.

This first known qualitative study evaluating real-life patient and caregiver burden showed that nmDMD has a substantial and multi-faceted impact on both patient and caregivers, including physical, emotional, social and time related consequences.^{4,5,6} As part of the research, several caregivers reported a positive change in their child's condition following initiation with Translarna™ (ataluren) and subsequently an improvement in health-related quality of life (HRQoL).^{4,5}

"These findings reinforce the need for treatments that address the high unmet need in Duchenne muscular dystrophy and the vital spill over benefits that treatments could also provide for families and caregivers. By improving symptoms, reducing loss of function, or delaying disease progression, treatments have the potential to positively impact the health-related quality of life of individuals, families and caregivers living with Duchenne," said Sarah Acaster, Principal Investigator, Acaster Lloyd Consulting Ltd (ALC).

Overview of PTC data presented at ISPOR EU 2020

Title	Abstracts
AADC deficiency	
Age at onset and frequency of clinical signs and symptoms in patients with AADC Deficiency: A systematic literature review	PRO8
Identifying appropriate PROXY diseases for estimating long-term survival and impact on health-related quality of life of patients with Aromatic l-amino acid decarboxylase deficiency (AADC-d)	PRO10
Estimating the long-term effects of therapy for patients with Aromatic l-amino acid decarboxylase deficiency (AADC-d) using individual participant data	PRO11
Deriving vignettes for a rare disease using parent, caregiver and clinician interviews to evaluate the impact on health-related quality of life	PRO118
A vignette study to derive health state utilities for Aromatic l-amino acid decarboxylase (AADC) deficiency in the United Kingdom (UK)	PRO127
A discrete choice experiment to derive health state utilities for Aromatic l-amino acid decarboxylase (AADC) deficiency in the United Kingdom	PRO135

Validating vignettes for a rare disease using clinician interviews to evaluate the impact on health-related quality of life in Aromatic L-amino acid decarboxylase (AADC) deficiency in France	PRO133
Nonsense mutation Duchenne muscular dystrophy	
Symptoms and impacts of nonsense mutation Duchenne muscular dystrophy at different stages of ambulation	PRO136
The development of a conceptual model on the impact of caring for an individual with nonsense mutation Duchenne muscular dystrophy	PRO117
Symptoms and impacts of nonsense mutation Duchenne muscular dystrophy: A qualitative study and the development of a patient-centered conceptual model	PRO119
Disease progression indicators in Duchenne muscular dystrophy for use in matched analyses comparing treatment effects: a targeted literature review	PRO2

About aromatic L-amino acid decarboxylase deficiency (AADC-d)

AADC-d is an ultra-rare, genetic, life-shortening disorder that causes severe disability and ongoing physical and mental suffering from the first few months of life, with many reliant on feeding tubes or breathing support to survive, and life-long, 24-hour care.¹ AADC deficiency leads to 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.¹ Significant health issues include infections, orthopedic and cardiac complications, and other comorbidities.

About Duchenne Muscular Dystrophy

Primarily affecting males, Duchenne muscular dystrophy (Duchenne) is a rare and fatal genetic disorder that results in progressive muscle weakness from early childhood and leads to premature death in the mid-twenties due to heart and respiratory failure. It is a progressive muscle disorder caused by the lack of functional dystrophin protein. Dystrophin is critical to the structural stability of all muscles, including skeletal, diaphragm, and heart muscles. Patients with Duchenne can lose the ability to walk as early as age ten, followed by loss of the use of their arms. Duchenne patients subsequently experience life-threatening lung complications, requiring the need for ventilation support, and heart complications in their late teens and twenties.

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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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 regulatory submissions and responses and other matters; expectations with respect to PTC's gene therapy platform; 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: expectations with respect to PTC's gene therapy platform; 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; 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 Translarna, Emflaza or 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.

* Six symptom domains of AADC deficiency: Developmental delays, movement disorders (e.g. oculogyric crises, muscle spasms), problems with muscle tone (e.g. floppiness), behavior and sleeping problems, autonomic function problems (e.g. problems with the regulation on involuntary body processes such as heart function and temperature control), and cardiovascular/gastrointestinal/endocrine/respiratory problems.

- ¹ Wassenberg. T, et al. Consensus guideline for the diagnosis and treatment of aromatic l-amino acid decarboxylase (AADC) deficiency. *Orphanet J Rare Dis.* 2017;12:12.
- ² BakidouA, Werner C, Buesch K. *Age at onset and frequency of clinical signs and symptoms in patients with AADC deficiency: A systematic literature review.* Poster to be presented at the ISPOR Europe Annual Meeting; November 16-19, 2020.
- ³ Simons C, Sudhapalli P, Buesch K, Bennison C. *Estimating the long-term effects of therapy for patients with aromatic l-amino acid decarboxylase deficiency (AADC-d) using individual participant data.* Poster to be presented at the ISPOR Europe Annual Meeting; November 16-19, 2020.
- ⁴ Williams K, Buesch K, PiglowskaN, Davidson I, RanceM, BoehnkeA, AcasterS. *The development of a conceptual model on the impact of caring for an individual with nonsense mutation Duchenne Muscular Dystrophy.* Poster to be presented at the ISPOR Europe Annual Meeting; November 16-19, 2020.
- ⁵ Williams K, Buesch K, PiglowskaN, AcasterS. *Symptoms and impacts of nonsense mutation Duchenne Muscular Dystrophy: A qualitative study and the development of a patient-centredconceptual model.* Poster to be presented at the ISPOR Europe Annual Meeting; November 16-19, 2020.
- ⁶ Williams K, Buesch K, Piglowska N, Davison I, Rance M, Boehnke A, Acaster S. *Symptoms and impacts of nonsense mutation Duchenne Muscular Dystrophy at different stages of ambulation.* Poster to be presented at the ISPOR Europe Annual Meeting; November 16-19, 2020
- ⁷ Van Riuten H, Bushby K, Guglieri M. State-Of-The-Art Advances in Duchenne Muscular Dystrophy. *EMJ.* 2017;2[1]:90-99.

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