PTC Therapeutics Presents New Sepiapterin Data from Ongoing Studies

March 20, 2025

—Results show 97% of phenylalanine (Phe) tolerance study participants were able to increase their dietary Phe intake with a mean increase of 126% —

— Genetic variant analysis of Phase 3 APHENITY trial demonstrates meaningful effect in classical subjects with non-BH4 responsive genotypes —

WARREN, N.J., March 20, 2025 /PRNewswire/ -- PTC Therapeutics, Inc. (NASDAQ: PTCT) today shared new data being presented from the Phase 3 APHENITY trial and subsequent open-label extension study at the 2025 American College of Medical Genetics and Genomics (ACMG) Annual Clinical Genetics Meeting. These data provide further evidence of the potential meaningful benefits of sepiapterin treatment for the full spectrum of phenylketonuria (PKU) patients, including significant diet liberalization.

Key data presented include:

- Over 97% of subjects participating in the Phe tolerance protocol of the APHENITY open-label extension study demonstrate the ability to liberalize their diet while on sepiapterin treatment, with a mean increase in protein intake of 126%
- 66% of subjects participating in the Phe tolerance sub study reached or exceeded the age-adjusted recommended daily allowance of protein intake for an individual without PKU, while maintaining control of blood Phe levels
- Genetic variant analysis of subjects participating in the APHENITY study demonstrates that over 70% had a Genotype-Phenotype Value (GPV) consistent with classical PKU

"These new data provide further evidence of the meaningful benefit sepiapterin can provide for children and adults with PKU, including those with the most severe form of the disease," said Matthew B. Klein, M.D., Chief Executive Officer of PTC Therapeutics. "Furthermore, the treatment benefit demonstrated in individuals with mutations not responsive to BH4 and consistent with classical disease phenotype support the potential to penetrate all key patient segments."

The data were presented in a poster titled "Interim Results From the APHENITY Extension Study: Sepiapterin Reduces Blood Phe with Improved Dietary Phe Tolerance in Participants With Phenylketonuria" and in a symposium presented by Nicola Longo, M.D., Ph.D., Professor and Chief of Division of Clinical Genetics at the University of California, Los Angeles and Suzanne Hollander, M.S., R.D., L.D.N., Senior Clinical Nutrition Specialist and Associate Director of PKU Program at Boston Children's Hospital.

About Sepiapterin
Sepiapterin (formerly PTC923), an oral formulation of synthetic sepiapterin, has a dual mechanism of action to increase activity of the phenylalanine hydroxylase (PAH) enzyme. First, sepiapterin is a precursor compound that is rapidly absorbed and converted intracellularly to tetrahydrobiopterin (BH4), a critical cofactor of PAH. Sepiapterin also has an independent pharmacological chaperone effect, correcting PAH misfolding to enhance the enzyme function. Through this dual mechanism of action, sepiapterin effectively reduces blood phenylalanine (Phe) levels and has the potential to treat a broad range of PKU patients.

About Phenylketonuria
Phenylketonuria (PKU) is a rare, inherited metabolic disease, which affects the brain. It is caused by a defect in the gene that helps create the enzyme needed to break down phenylalanine. If left untreated or poorly managed, phenylalanine – an essential amino acid found in all proteins and most foods – can build up to harmful levels in the body. This causes severe and irreversible disabilities, such as permanent intellectual disability, seizures, delayed development, memory loss, and behavioral and emotional problems. Newborns with phenylketonuria initially don't have any symptoms, but symptoms are usually progressive, and damage caused by toxic levels of phenylalanine in the first few years of life is irreversible. Diagnosis of phenylketonuria usually takes place during newborn screening programs. There are an estimated 58,000 people with phenylketonuria globally.

About PTC Therapeutics, Inc.
PTC is a global biopharmaceutical company focused on the discovery, development and commercialization of clinically differentiated medicines that provide benefits to children and adults living 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, X, and LinkedIn.

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Forward-Looking Statement
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, commercialization and other matters with respect to its products and product candidates; PTC's strategy, future operations, future financial position, future revenues, projected costs; the extent, timing and financial aspects of our strategic pipeline prioritization and reductions in workforce; 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 sepiapterin, including any regulatory submissions and potential approvals, commercialization, the potential achievement of 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 sepiapterin.

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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