PTC Therapeutics in Gene Therapy

From Lab to Patient

What causes genetic disorders?

Genes are small sections of DNA that provide instructions for the body to make proteins involved in processes essential for life.¹



A change or fault in the gene, called a mutation, may cause a 'genetic disorder'.²



Gene therapy: new hope for previously intractable disease.

Gene therapy aims to treat the genetic disorders by replacing or inactivating genes or introducing corrective genes into cells.³

New genes are transferred into cells using transporters known as vectors; made from modified, inactivated viruses that are not harmful.³

In-vivo – directly infused inside the patient³

Ex-vivo – transferred outside the body and then returned to the patient³

By correcting the genetic cause of disease, a single dose of a gene therapy can offer benefits lasting years, if not a lifetime, versus a lifetime of ongoing treatment.⁴ Gene therapy is bringing new hope for thousands of people, many of whom are children with disabling and life-limiting conditions.⁵

Gene therapy holds tremendous promise for some of the most debilitating genetic disorders. PTC Therapeutics is at the forefront of this exciting and transformative area.



Rare Resolve for Rare Disease – an ongoing campaign to raise awareness of AADC deficiency and accelerate the recognition, referral and diagnosis of patients with this rare disease



2019

Support for no-cost access to 3-OMD screening tests for patients suspected of AADC deficiency and biobank screening initiatives

Masterclass webinars for specialists worldwide

2020

PTC Therapeutics seeks regulatory approval for PTC-AADC for AADC deficiency in the EU



Surgeons from the US and Germany observed the treatment in Taiwan



2019

PTC Therapeutics starts working with select treatment centers across the US and Europe to establish a standard of care for the delivery of its AADC deficiency gene therapy



2020

PTC Therapeutics launches a variety of no cost genetic and blood testing options globally, for HCPs to test patients suspected of having AADC deficiency



CHMP reviewing marketing access application for PTC-AADC for patients with AADC deficiency



2021

PTC Therapeutics plans to submit Biologics License Application for AADC deficiency gene therapy to the FDA by year end



PTC Therapeutics has one of the most advanced gene therapy pipelines for rare neurological disorders*

PTC-AADC for AADC deficiency (DDC gene)

AADC deficiency typically causes severe disability and suffering from the first months after birth, affecting every aspect of life – physical, mental and behavioural⁸⁻¹²

PTC-FA for Friedreich ataxia (FXN gene)

Friedreich ataxia progressively robs patients of their ability to walk, speak, see and hear¹³

PTC-AS for Angelman syndrome (UBE3A gene)

Angelman syndrome is characterized by profound intellectual and developmental delays¹⁴

* Information last updated October 2021

Abbreviations

AADC, Aromatic L-amino acid decarboxylase; HCP, Healthcare professional; 3-OMD, 3-O-methyldopa

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