Horizon Therapeutics plc and Invitae Corporation Launch Urea Cycle Disorder Genetic Testing Program

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-- New program improves access and speed to receiving a diagnosis for people and extended family members who may be living with Urea Cycle Disorders --

DUBLIN--(BUSINESS WIRE)--May 29, 2019-- Horizon Therapeutics plc (Nasdaq: HZNP) in partnership with Invitae Corporation (NYSE: NVTA) today announced a new urea cycle disorder (UCD) genetic testing program designed to speed diagnosis and enable physicians to offer genetic testing at no-charge to people who may have a UCD, including their family members. Horizon's support of the UCD Genetic Testing Program facilitates the genetic tests and services offered and performed by Invitae.

This press release features multimedia. View the full release here: https://www.businesswire.com/news/home/20190529005164/en/

UCDs are rare, genetic disorders characterized by potentially toxic elevations of ammonia levels in the blood and brain (hyperammonemia). UCD symptoms may first occur at any age depending on the severity of the disorder, with more severe defects presenting earlier in life.[1]

"Patients with urea cycle disorders, particularly those who do not develop severe clinical signs of overwhelming hyperammonemia in the newborn period, can face a long and frustrating path to a correct diagnosis. Current diagnostic paradigms are complex and involve measurement of a variety of blood and urine substances – and still may not yield a definitive diagnosis" said Robert Nussbaum, M.D., chief medical officer, Invitae. "Earlier access to genetic testing contributes to making an accurate diagnosis so that the clinicians treating these patients can turn more quickly to effective treatments and counseling. One prime example are mothers with genetic variants associated with the most common form of UCD, a deficiency related to the *OTC* gene, who often go undiagnosed because they only have vague, episodic symptoms and are unaware that they themselves are at risk for hyperammonemia or are at risk for passing the disorder on to their children. We're pleased to be working with Horizon on this important program to get more UCD patients access to the testing they need."

The UCD Genetic Testing Program utilizes the Invitae Hyperammonemia Panel to evaluate 58 genes that are associated with the enzymes and transporter proteins responsible for the production and detoxification of ammonia. The most common type of UCD, which is caused by changes in the *OTC* gene, can be passed on from a mother to her child via the X chromosome. Very rarely, *OTC* variants can be passed down from a father to his daughter. If a parent with an *OTC* genetic variant is a carrier, they may not know until their child is diagnosed. A carrier can also be affected by the disease without knowing it, and may not experience symptoms until later in life.[1]

"We have learned that many UCD families still do not have access to genetic testing," said Jeffrey Kent, M.D., senior vice president, medical affairs, Horizon. "As part of our long-standing commitment to the UCD community, we strive to create solutions to enhance the lives of all impacted families. This partnership with Invitae will reduce current barriers and facilitate improved access to genetic testing for families with UCDs."

To learn more about the UCD Genetic Testing Program, visit the Invitae website.

Horizon provides financial support to Invitae for this program, which enables physicians to offer genetic testing at no-charge to their patients. Patient privacy is strictly protected and Horizon does not have any access to any identifiable patient information. Tests and services are performed by Invitae. Healthcare professionals must confirm that patients meet certain criteria to use the program. Genetic testing and counseling are only available in the United States.

About Urea Cycle Disorder

A UCD is a rare genetic disorder that affects approximately 1 in 35,000 live births in the United States. It is caused by an enzyme deficiency in the urea cycle, a process that is responsible for converting excess ammonia from the bloodstream and ultimately removing it from the body. Because of this, people with a UCD experience hyperammonemia, or elevated ammonia levels in their blood, that can then reach the brain and cause irreversible brain damage, coma or death.

About Invitae

Invitae Corporation is a leading advanced medical genetics company, whose mission is to bring comprehensive genetic information into mainstream medicine to improve healthcare for billions of people. Invitae's goal is to aggregate the world's genetic tests into a single service with higher quality, faster turnaround time, and lower prices. For more information, visit the company's website at invitae.com.

About Horizon

Horizon is focused on researching, developing and commercializing medicines that address critical needs for people impacted by rare and rheumatic diseases. Our pipeline is purposeful: we apply scientific expertise and courage to bring clinically meaningful therapies to patients. We believe science and compassion must work together to transform lives. For more information on how we go to incredible lengths to impact lives, please visit www.horizontherapeutics.com, follow us @HorizonNews on Twitter, like us on Facebook or explore career opportunities on LinkedIn.

[1] Ornithine transcarbamylase deficiency. National Organizations for Rare Disorders (NORD) website. https://rarediseases.org/rare-diseases/ornithine-transcarbamylase-deficiency. Accessed May 13, 2019.

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