

## Pharming Group, in collaboration with Invitae Corporation, launches genetic testing program ‘navigateAPDS’ in US and Canada

- **New program expected to improve access to genetic testing for activated PI3K delta syndrome (APDS), an ultra-rare immunodeficiency disease**
- **Program has potential to advance clinical research in APDS as a result of earlier diagnosis**

**Leiden, The Netherlands, 2 March 2021:** Pharming Group N.V. (“Pharming” or “the Company”) (Euronext Amsterdam: PHARM/Nasdaq: PHAR), a global, commercial stage biopharmaceutical company developing innovative protein replacement therapies and precision medicines for the treatment of rare diseases and unmet medical needs, in collaboration with Invitae Corporation (NYSE: NVTA, “Invitae”), a leading medical genetics company, announces the launch of a sponsored genetic testing program, navigateAPDS, designed to assist clinicians in identifying patients and their family members with activated PI3K delta syndrome (APDS), which may lead to earlier diagnosis.

APDS is an ultra-rare primary immunodeficiency disease caused by a genetic mutation affects approximately 1-2 people per million. Patients are often misdiagnosed with other immunodeficiencies or autoimmune disorders and often have a protracted course to obtain a correct diagnosis. A definitive diagnosis can be made only by a genetic test. Current treatment is generally limited to supportive therapies such as antibiotics and the use of immunoglobulin replacement therapy. There is no approved therapy for the treatment of APDS, however, clinical trials are currently ongoing, including Pharming’s pivotal-stage development program for leniolisib, a small molecule phosphoinositide 3-kinase delta (PI3K $\delta$ ) inhibitor, under development by Novartis and Pharming to treat patients with APDS.

Pharming’s support of the program will facilitate genetic testing and counselling for eligible individuals in the United States and Canada at no charge. NavigateAPDS will use the Invitae Primary Immunodeficiency Panel (PI), which analyzes up to 407 genes that are associated with inherited disorders of the immune system. In addition to providing genetic testing to individuals who may present with a clinical picture known to be associated with APDS, navigateAPDS will offer pre-test and post-test genetic counseling through a third party, and all blood relatives of patients found to have a P/LP variant for APDS are qualified to be tested through the program. By offering access to the full PI panel, physicians and patients are more likely to identify the underlying cause and potential diagnosis without the need for additional expanded patient testing.

**Sijmen de Vries, Chief Executive Officer commented:**

*"Our partnership with Invitae is an important step towards simplifying access to testing, which may allow for an expedited and accurate diagnosis for patients suffering from a primary immunodeficiency such as APDS. Earlier diagnosis can favorably impact disease management and could have a positive effect on long-term outcomes and patients' quality of life. The program is also a key component in advancing clinical research by identifying the underlying causes of APDS, which will help us better understand these patients' potential to respond to investigational precision medicines."*

**Robert Nussbaum, M.D., Chief Medical Officer of Invitae commented:**

*"Genetic information is a powerful tool that can improve outcomes for patients and their families, both by supporting more rapid diagnosis of ultra-rare diseases such as APDS and also by enabling access to clinical trials. Partnerships like our work with Pharming may enable the clinical trials necessary to develop precision therapies in areas of significant unmet need."*

To learn more about the navigateAPDS program, visit [www.invitae.com/navigateapds](http://www.invitae.com/navigateapds).

## **About APDS**

APDS is an ultra-rare primary immunodeficiency first fully described in 2013 that affects approximately 1-2 people per million. APDS occurs when there is an abnormal change in either one of two specific genes, the PIK3CD gene or the PIK3R1 gene. The genes follow an autosomal dominant mode of inheritance which means one copy of the altered gene from either biological parent is sufficient to cause the disorder. The genes are involved in making parts of a protein that helps in the growth and division of white blood cells, particularly the B-cell and T-cell lymphocytes. APDS is present at birth and signs and symptoms start early in childhood with affected individuals having increased susceptibility to a myriad of inflammatory conditions including recurrent severe respiratory tract infections, chronic benign lymphoproliferation, hematopoietic malignancies, and/or autoimmune diseases. Patients are often misdiagnosed with other immunodeficiencies or autoimmune disorders and often have a protracted course to obtain a correct diagnosis. A definitive diagnosis can only be made by a genetic test and, once appropriately identified, regular specialized health check-ups and customized treatment plans are required. Current treatment is generally limited to supportive therapies such as antibiotics and the use of immunoglobulin replacement therapy. There is no approved therapy for the treatment of APDS, however, clinical trials are currently ongoing.

To learn more, visit [www.allaboutapds.com](http://www.allaboutapds.com).

## **About Pharming Group N.V.**

Pharming Group N.V. is a global, commercial stage biopharmaceutical company developing innovative protein replacement therapies and precision medicines for the treatment of rare diseases and unmet medical needs.

The flagship of our portfolio is our recombinant human C1 esterase inhibitor, or rhC1INH, franchise. C1INH is a naturally occurring protein that downregulates the complement cascade in order to control swelling in affected tissues.

Our lead product, RUCONEST® is the first and only plasma-free rhC1INH protein replacement therapy. It is approved for the treatment of acute hereditary angioedema, or HAE, attacks. We are commercializing RUCONEST® in the United States, the European Union and the United Kingdom through our own sales and marketing organization, and the rest of the world through our distribution network.

We are also developing rhC1INH for subsequent indications, including pre-eclampsia, acute kidney injury and we also investigating the clinical efficacy of rhC1INH in COVID-19.

In addition, we are studying our oral precision medicine, leniolisib (a phosphoinositide 3-kinase delta, or PI3K delta, inhibitor), for the treatment of activated PI3K delta syndrome, or APDS, in a registration enabling Phase 2/3 study in the United States and Europe.

Furthermore, we are also leveraging our transgenic manufacturing technology to develop next-generation protein replacement therapies most notably for Pompe disease, which program is currently in the preclinical stage.

For more information please visit the company's website: [www.pharming.com](http://www.pharming.com).

## About Invitae

Invitae Corporation (NYSE: NVTA) is a leading 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 [www.invitae.com](http://www.invitae.com).

## Forward-looking Statements

*This press release contains forward-looking statements, including with respect to timing and progress of Pharming's preclinical studies and clinical trials of its product candidates, Pharming's clinical and commercial prospects, Pharming's ability to overcome the challenges posed by the COVID-19 pandemic to the conduct of its business, and Pharming's expectations regarding its*

*projected working capital requirements and cash resources, which statements are subject to a number of risks, uncertainties and assumptions, including, but not limited to the scope, progress and expansion of Pharming's clinical trials and ramifications for the cost thereof; and clinical, scientific, regulatory and technical developments. In light of these risks and uncertainties, and other risks and uncertainties that are described in Pharming's 2019 Annual Report and its report for the nine months ended 30 September 2020, the events and circumstances discussed in such forward-looking statements may not occur, and Pharming's actual results could differ materially and adversely from those anticipated or implied thereby. Any forward-looking statements speak only as of the date of this press release and are based on information available to Pharming as of the date of this release.*

### **Inside Information**

*This press release relates to the disclosure of information that qualifies, or may have qualified, as inside information within the meaning of Article 7(1) of the EU Market Abuse Regulation.*

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