X4 Pharmaceuticals and Invitae Announce Partnership to Provide No-Cost Genetic Testing to Patients Suspected of Primary Immunodeficiency Disease

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Genetic testing to enable improved patient diagnoses and provide valuable disease-specific genetic insights for X4 clinical development of novel therapies

CAMBRIDGE, Mass.--(BUSINESS WIRE)--Jun. 12, 2019-- X4 Pharmaceuticals, Inc. (Nasdaq: XFOR), a clinical-stage biopharmaceutical company focused on the development of novel therapeutics for the treatment of rare diseases, and Invitae Corporation (NYSE: NVTA), a leader in medical genetics, today announced a partnership to provide genetic testing at no cost to patients through its collaborative PATH4WARD program. This initiative provides greater access to faster and earlier diagnosis for individuals who may carry a genetic mutation known to be associated with WHIM syndrome and Severe Congenital Neutropenia (SCN) – a group of rare inherited primary immunodeficiencies (PIs).

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“Rare diseases such as WHIM and SCN, don’t often receive the attention and research that patients and their families deserve in order to discover and develop new therapeutic options,” said Paula Ragan, Ph.D., President and Chief Executive Officer of X4 Pharmaceuticals. “Working with a strong and committed partner like Invitae allows us to facilitate quick and accurate diagnoses for patients, which can have a profound impact on their disease management and overall quality of life. Simultaneously, this partnership provides X4 with a clear path to gather critical data to identify the underlying genetic causes of PIs and deepen our understanding of these patients’ potential to respond to novel investigational therapies.”

In addition to providing genetic testing to individuals who may present with a clinical picture known to be associated with WHIM or SCN, PATH4WARD will offer genetic counseling, as well as family variant testing (FVT) for all blood relatives of patients found to have a pathogenic or likely pathogenic variant at no additional charge. If the initial testing does not show mutations associated with WHIM or SCN, physicians will be able to access a broader PI panel through the program for expanded patient testing.

“Time-to-diagnosis can mean all the difference to people living with rare disease, and too often these families find themselves in diagnostic odysseys that can last years,” said Robert Nussbaum, M.D., Chief Medical Officer of Invitae. “We are thrilled to partner with X4 Pharmaceuticals to increase access to genetic testing that can help diagnose more patients more quickly, thereby identifying more patients eligible for precision therapies and deepening the clinical community’s understanding of PI disorders such as WHIM and SCN.”

About Severe Congenital Neutropenia

Severe congenital neutropenia (SCN) comprises a group of rare hematological diseases characterized by impaired maturation of white blood cells. Affecting an estimated 3 to 8.5 cases per one million individuals, patients with SCN are prone to recurrent, often life-threatening infections beginning in their first months of life.[1]

About WHIM Syndrome

WHIM syndrome is a primary immunodeficiency disease caused by genetic mutations in the CXCR4 receptor gene resulting in susceptibility to certain types of infections. WHIM is an abbreviation for the characteristic clinical symptoms of the syndrome: Warts, Hypogammaglobulinemia, Infections, and Myelokathexis. Within the overall category of primary immunodeficiencies, there are between 15,000 and 100,000 patients in the U.S. who are classified with primary immunodeficiency disease of unknown origin – of which WHIM is one.[2,3,4] WHIM is a rare disorder and the precise prevalence or incidence of patients that have the genetic mutation responsible for WHIM is unknown. Individuals with WHIM are more susceptible to potentially life-threatening bacterial infections.[5] Additionally, WHIM is associated with significant morbidity beginning in early childhood and continuing throughout life. Current therapy is limited to treatment of acute infections with antibiotics or prevention through the use of intravenous immunoglobulin or G-CSF. There is no approved therapy for the treatment of WHIM.

About X4 Pharmaceuticals

X4 Pharmaceuticals is developing novel therapeutics designed to improve immune cell trafficking to treat rare diseases, including primary immunodeficiencies and certain cancers. X4’s oral small molecule drug candidates antagonize the CXCR4 pathway, which plays a central role in immune surveillance. X4’s most advanced product candidate, mavorixafor (X4P-001), is expected to commence a global Phase 3 pivotal trial in patients with WHIM syndrome, a rare genetic, primary immunodeficiency disease, in the second quarter of 2019 and is currently also under investigation in a Phase 2a clinical trial in clear cell renal cell carcinoma. X4 was founded and is led by a team with extensive product development and commercialization expertise, including several former members of the Genzyme leadership team, and is located in Cambridge, Massachusetts. For more information, please visit www.x4pharma.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 invitae.com.

Forward-Looking Statements

This press release contains forward-looking statements within the meaning of the Private Securities Litigation Reform Act of 1995, as amended. The words “may,” “will,” “could,” “would,” “should,” “expect,” “plan,” “anticipate,” “intend,” “believe,” “estimate,” “predict,” “project,” “potential,” “continue,” “target” and similar expressions are intended to identify forward-looking statements, although not all forward-looking statements contain these identifying words. Forward-looking statements include statements regarding plans for, or progress, scope, cost, duration or results or timing for the initiation, completion or availability of results of development of mavorixafor (X4P-001) or any of our other product candidates or programs, including regarding the Phase 3 clinical trial of mavorixafor for the treatment of patients with WHIM syndrome, the target indication(s) for development, the size, design, population, location, conduct, objective, duration or endpoints of any clinical trial, or the timing for initiation or completion of or reporting of results from any clinical trial, the potential benefits of mavorixafor, or any other product candidate or program or the commercial opportunity in any target indication as well as the expected offerings and benefits of the PATH4WARD program and X4’s relationship with Invitae. These statements are subject to various risks and uncertainties, actual results could differ materially from those projected and X4 cautions investors not to place undue reliance on the forward-looking statements in this press release. These risks and uncertainties include, without limitation, the risk that trials and studies may be delayed and may not have satisfactory outcomes, potential adverse effects arising from the testing or use of mavorixafor or other product candidates, the risk that costs required to develop mavorixafor or other product candidates or to expand our operations will be higher than anticipated and the risk that the PATH4WARD program and X4’s relationship with Invitae will not be successful. Any forward-looking statements in this press release are based on management's current expectations and beliefs and are subject to a number of risks, uncertainties and important factors that may cause actual events or results to differ materially from those expressed or implied by any forward-looking statements contained in this press release, including, without limitation, the risks and uncertainties described in the section entitled “Risk Factors” in X4’s most recent Annual Report on Form 10-K filed with the Securities and Exchange Commission (SEC), as updated by X4’s Current Report on Form 8-K filed with the SEC on April 11, 2019, and in other filings X4 makes with the SEC from time to time. X4 undertakes no obligation to update the information contained in this press release to reflect subsequently occurring events or circumstances.

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[1] Welte et al., Nature reviews, 2017.

[2] Boyle JM, Buckley, RH, Population Prevalence of Diagnosed Primary Immunodeficiency Diseases in the United States. J Clin Immunol 2007;27:497–502.

[3] Gathmann B, Grimbacher B, et al. The European internet-based patient and research database for primary immunodeficiencies: results 2006–2008. Clin Exp Immunol. 2009 Sep;157 Suppl 1:3-11.

[4] Modell V, Gee B, et al. Global study of primary immunodeficiency diseases (PI) — diagnosis, treatment, and economic impact: an updated report from the Jeffrey Modell Foundation. Immunol Res. 2011;51:61–70.

[5]Primary Immunodeficiency Foundation: https://primaryimmune.org/disease/whim-syndrome.

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Source: X4 Pharmaceuticals, Inc.

Investors:
Stephanie Carrington
Westwicke, an ICR company
646-277-1282
Stephanie.Carrington@icrinc.com

Media:
Darcie Robinson
Westwicke, an ICR company
203-919-7905
Darcie.robinson@icrinc.com