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Study aims to establish a genetic screening approach to aid in diagnosis of patients with WHIM syndrome

NEW YORK, N.Y. and CAMBRIDGE, Mass. April 24, 2018 – The Jeffrey Modell Foundation, a non-profit organization for primary immunodeficiency diseases, and X4 Pharmaceuticals, a clinical stage biotechnology company developing novel CXCR4 inhibitor drugs, today announced that they are jointly sponsoring a clinical study establishing a genetic screening protocol to help identify patients who have WHIM syndrome (WHIM), a rare, chronic and life-threatening primary immunodeficiency.

WHIM is caused by a mutation in a patient's C-X-C chemokine receptor type 4 (CXCR4) genes. The objective of the collaborative study is to establish a systematic diagnostic approach for WHIM by combining clinical features and genetic testing. Currently WHIM patients are seen by a diverse group of medical specialists and often go undiagnosed or are broadly classified as patients with primary immunodeficiency disease of unknown origin and/or unconfirmed diagnosis.

"The journey for patients with WHIM is long and winding, and can take them to numerous pediatric or adult specialists before specific genetic diagnosis and treatment may be offered," said Dr. Jolan Walter, MD, PhD, Division Chief and Robert A. Good Endowed Chair of Pediatric Allergy Immunology, University of South Florida at Johns Hopkins All Children's Hospital and lead investigator of the study. "This study aims to establish a systematic approach for the early diagnosis of WHIM patients among physicians working in different fields. The design allows for targeted genetic screening for mutated CXCR4 in WHIM patients at no cost to patients or insurers."

Through the collaborative clinical study, up to 300 patients will be evaluated with genetic screening for WHIM. The genetic testing will be carried out by PreventionGenetics, a leading, CLIA and ISO 15189:2012 accredited clinical DNA testing laboratory.

"WHIM syndrome is one of the primary immunodeficiency diseases which is probably much more common than is currently recognized. Linking clinical presentation and genetic diagnosis will help physicians to care for this underserved patient population. Early diagnosis could lead to prevention of some of the chronic consequences of WHIM such as deafness, chronic lung disease and invasive HPV related cancers," said Dr. David C. Dale, MD, Professor of Medicine and former Dean of the School of Medicine at the University of Washington Medical Center.

"Developing diagnostic protocols that use genetic testing is essential for advancing therapeutic innovation to treat primary immunodeficiencies," said Vicki Modell, Co-Founder of Jeffrey Modell Foundation. "This collaboration is an example of the power of public-private partnerships to drive towards a better understanding of a particular primary immunodeficiency with the goal of efficient early identification to improve patient outcomes."

"Through this collaboration, we will evaluate up to 300 patients, representing the largest WHIM study to date," said Sudha Parasuraman, MD, Chief Medical Officer of X4 Pharmaceuticals. "X4 is pleased to be partnering with the Jeffrey Modell Foundation on this crucial step forward in WHIM patient care. Identification through genetic screening is key to shortening a WHIM patient's path to diagnosis and will hopefully enable them to access more effective treatments in the future."

About WHIM Syndrome

WHIM syndrome is a primary immunodeficiency disease caused by genetic mutations in the CXCR4 receptor generesulting 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 US that are classified with primary immunodeficiency disease of unknown origin – of which WHIM is one.[1,2,3] WHIM syndrome is a rare disorder and the precise prevalence or incidence of patients that have the genetic mutation responsible for WHIM syndrome is unknown. Because patients are highly susceptible to infections, WHIM syndrome 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 syndrome.

About Jeffrey Modell Foundation

Vicki and Fred Modell established the Jeffrey Modell Foundation (JMF) in 1987, in memory of their son Jeffrey, who died at the age of fifteen from complications of Primary Immunodeficiency – a genetic condition that is chronic, serious, and often fatal. JMF is a global nonprofit organization dedicated to early diagnosis, meaningful treatments and, ultimately, cures through research, physician education, public awareness, advocacy, patient support, and newborn screening. The Jeffrey Modell Centers Network includes 781 physicians at 356 academic institutions, in 273 cities, 86 countries, spanning 6 continents and growing. For more information about PI, visit www.info4pi.org or email the Jeffrey Modell Foundation at info4pi@jmfworld.org.

About PreventionGenetics

Founded in 2004 and located in Marshfield, Wisconsin, PreventionGenetics is a CLIA and ISO 15189:2012 accredited clinical DNA testing laboratory. PreventionGenetics provides patients with sequencing and deletion/duplication tests for nearly all clinically relevant genes. These tests include our powerful and comprehensive whole exome sequencing test, PGxomeTM.

About X4 Pharmaceuticals

X4 Pharmaceuticals is developing novel therapeutics designed to improve immune cell trafficking to treat cancer and rare diseases. The Company's oral small molecule drug candidates inhibit the CXCR4 receptor, a pathway which plays a central role in immune surveillance. X4's most advanced product candidate, X4P-001-RD, is in a Phase 2/3 study in patients with WHIM syndrome, a rare genetic, primary immunodeficiency disease. X4P-001-IO is currently under investigation in multiple Phase 1/2 studies in refractory clear cell renal cell carcinoma (ccRCC) and melanoma. X4 was founded and is led by a team with deep product development and commercialization expertise, including several former members of the Genzyme leadership team, and is located in Cambridge, MA. For more information, visit x4.theyatesnetwork.com.

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

[2] 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.

[3] 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.

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