Researchers Find Genetic Link to Rare, Life-Threatening Lung Condition

SALT LAKE CITY , UT, USA , May 29, 2018 /EINPresswire.com/ -- Chronic thromboembolic pulmonary hypertension (CTEPH), which is a rare but deadly condition that can occur after initial treatment of a blood clot in the lung, may be an inherited genetic disease, according to a first-of-its-kind study from researchers at Intermountain Healthcare in Salt Lake City.

Chronic thromboembolic pulmonary hypertension (CTEPH), which is a rare but deadly condition that can occur after initial treatment of a blood clot in the lung, may be an inherited genetic disease, according to new study.

When clinicians treat a pulmonary embolism, or blood clot in the lungs, they use blood thinners to break up the clot and reopen the affected artery. However, in patients with CTEPH, the blood clots don't dissolve, which leads to a rise in blood pressure and strain on the heart as it tries to pump blood through the still-blocked arteries in the lungs.

"Until now, no genetic link has been identified to indicate CTEPH is hereditary," said Mark Dodson, MD, PhD, medical director of the chronic thromboembolic pulmonary hypertension program at Intermountain Medical Center. "Our research suggests that CTEPH may in fact be a genetic disease, with inherited genetic risk factors passed down from one generation to the next."

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> *Dr. Mark Dodson, Intermountain Medical Center*

Results of the study were presented at the American Thoracic Society's annual conference this month in San Diego.

CTEPH is a long-term complication that occurs in some patients who've had a pulmonary embolism, which is a blood clot that typically starts in the legs, then travels to the lungs. Usually, patients with a pulmonary embolism have shortness of breath, low blood oxygen levels, chest pain, and/or lightheadedness.

Doctors treat pulmonary embolism with blood-thinning medication, which usually dissolves the blood clots over a period of months and prevents new clots from forming. For reasons that aren't well-understood, blood clots don't properly dissolve in patients with CTEPH, and so additional treatments are needed.

CTEPH occurs in about four percent of patients who've been diagnosed with a pulmonary

embolism, but can also occur in patients who've never had one. Up to 25 percent of patients diagnosed with CTEPH have no prior history of acute pulmonary embolism.

For the study, researchers reviewed medical records linked to the Utah Population Database to identify all patients who have ever been diagnosed with CTEPH in the stage of Utah. The Utah Population Database (UPDB) is a unique Utah resource that contains detailed genealogy records for approximately 4 million Utah residents dating back to the Mormon pioneers.

In many cases, genealogy data extends up to 15 generations deep. For this study, the researchers required that individuals included in the study have genealogy data in the UPDB that extends back at least three generations. The researchers then used the genealogy records in the UPDB to compare the average pair-wise relatedness of all possible pairs of patients with CTEPH to that of matched controls without CTEPH.

Researchers identified 141 Utah CTEPH cases, of which 66 met their criteria for adequacy of genealogy information. These 66 CTEPH cases were significantly more closely related than were controls, suggesting that the increased relatedness of CTEPH subjects is unlikely to have occurred by chance.

"The most important result of these findings is that we've identified a number of large pedigrees with multiple cases of CTEPH in the family," said Dr. Dodson. "These pedigrees are ideal tools to now use gene sequencing to identify the genes that cause CTEPH in these families."

Funding for the study funding was provided by donors to the Intermountain Research and Medical Foundation, which provides seed grant money for worthy research projects, such as this one, that lead to clinical applications.

Members of the research team included: Lynette Brown, MD, PhD; Gregory Elliott, MD; and Kristina Allen-Brady, PhD, from Intermountain Healthcare, and Lisa Cannon-Albright, PhD, from the University of Utah.

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