## G75 Molecular Genetic Testing in 323 Cases of Fatal Pulmonary Thromboembolism in the City of New York Revealed Racial Stratification

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The goal of this presentation is to investigate the frequency of these genetic risk factors in fatal PE and to understand the genotype and phenotype correlation.

This presentation will impact the forensic science community by presenting detailed characterization of the mutation spectrum in fatal PE is vital for providing accurate diagnosis of cause of death and efficient preventative treatment to the high-risk family members.

Fatal pulmonary thromboembolism (PE) is a common cause of death encountered in the forensic pathology setting and usually presents as a complication of deep venous thrombosis (DVT). The pathogenesis of venous thrombosis is multifactorial and requires interaction between both inherited and acquired risk factors. Heterozygous or homozygous Factor V Leiden (G1691A) or prothrombin (G20210A) mutations, and homozygous MTHFR (C677T) variant have been recognized as common independent genetic risk factors in DVT. In order to investigate the frequency of these genetic risk factors in fatal PE and to understand the genotype and phenotype correlation, we have validated a genetic testing method to detect the three common mutations.

Testing was conducted using multiplex PCR-SNaPshot technologies on postmortem tissue and blood samples. Between March 2005 and May 2007, we tested 323 fatal PE cases from the New York City Office of Chief Medical Examiner. The authors found that 48 of the 323 cases were positive for at least one mutation. The genetic testing results were categorized by the demographic data and acquired contributing factors. The overall frequency of three mutations in PE cases was found is highest in Whites (34.15 %), followed by Hispanics (28%), very low in Blacks (3%), and zero in Asians. In contrast, the number of fatal PE instances in our study is highest in Blacks (54.8%), followed by Whites (25.4%), and Hispanics (15.5%), and very rare in Asians (1.5%). Blacks were also associated with a high percentage of idiopathic PE with unknown acquired contributing factors. This study suggests that there are racial disparities in genetic risks contributing to fatal PE. In addition, comparing the incidences of PE in different races to the racial composition in New York City residents (44.7% Whites, 26.6% Blacks, and 9.8% of Asians), Blacks showed the highest incidences of fatal PE. Further research focused on delineating the genetic risks in black populations is warranted. Detailed characterization of the mutation spectrum in fatal PE is vital for providing accurate diagnosis of cause of death and efficient preventative treatment to the high-risk family members.

**Molecular Genetic Testing, Fatal Pulmonary Thromboembolism, Racial Stratification**

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