Research improves diagnosis and treatment of bleeding disorder

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A rare bleeding disorder that can lead to life-threatening bleeding episodes is misdiagnosed in 15 per cent of cases according to findings from a new international research project led by a Queen's professor.

"Correct diagnosis is critical because it determines the treatment decision," says Maha Othman, a professor in the Department of Anatomy and Cell Biology who led the three-year research project on the rare platelet type of von Willebrand disease (VWD).

Patients with VWD are commonly treated with drugs that help control their condition. However, these drugs aggravate bleeding in patients with the rarer platelet form of the disease, and misdiagnosis can leave these patients vulnerable to severe life-threatening bleeding episodes in situations like pregnancy and surgical operations.

Although both forms of VWD are genetic disorders that share many diagnostic features, the defect actually lies in two different genes. A correct diagnosis can only be made by closely examining certain areas of both genes to determine where the defect lies.

Dr. Othman's study is the first large study to investigate the occurrence of the two types of VWD worldwide and to evaluate DNA analysis as a diagnostic tool.

In addition to pioneering this international project, Dr. Othman has also created an online registry aimed at determining the frequency of the rare

platelet form of VWD and to collect data about the disorder. Despite its relative rarity, VWD is actually the most common genetically inherited bleeding disorder, affecting about one per cent of the general population.

Dr. Othman's research will be published in the March issue of the *Thrombosis and Haemostasis Journal*.

Provided by Queen's University