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The genetics of aortic valve disease

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A ortic valve disease represents a major health burden whose etiology remains poorly understood. Bicuspid aortic valve (BAV) is the most prevalent cardiac birth defects affecting 1% of the population. While present since birth, BAV is often diagnosed later in adulthood in the context of serious cardiovascular complications. These range from aortic rupture to calcified valve. As such BAV associated disease is degenerative and presently irreversible. Although BAV frequency is 1%, it is linked with the majority of valve replacement procedures in adults of all ages. The reason underlying the increased complications in individuals with BAV is not yet defined but may be due to geometry or genetics or both. We have used molecular biology and mouse genetics to discover BAV causing genes and the molecular basis for BAV associated disease. These basic discoveries are being translated for improved diagnosis and care of patients.

Biography

Mona Nemer is a Professor of Medicine and the Vice-President Research at the University of Otawa, the largest bilingual research-intensive university in the world. She has received her PhD from McGill University and has published over 150 papers in leading scientific journals. She serves on numerous national and international Advisory Boards and oversees the entire university research enterprise. She is a Fellow of the Royal Society of Canada, a Member of the Order of Canada and a Knight of the Order of Merit of the French Republic.

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