CSCC Award for Research Excellence
Sponsored by Ortho-Clinical Diagnostics

David E. C. Cole

*David Cole is a Medical Biochemist with a focus on biochemical and molecular genetics of inherite*d disorders. His current research interests include: familial ovarian cancer, osteoporosis and metabolic bone disease, and the physiology of human sulfur metabolism, especially homocysteine metabolism in vaso-occlusive disease. He is currently Professor in the Departments of Pathobiology, Medicine, and Paediatrics at the University of Toronto and a Senior Scientist of the University Health Network Research Institute. He is Director of the Adult Genetics Program at the University Health Network and a biochemist with the Toronto Medical Laboratories.

Dr. Cole got his BSc and MD from the University of Toronto in 1972 and 1975 respectively followed by a PhD from McGill. Subsequent to this, Dr Cole received his fellowships in the Canadian College of Medical Geneticists (Biochemical Genetics) in 1983, the Royal College of Physicians and Surgeons of Canada (Med. Biochem.) in 1989, the Royal College of Physicians and Surgeons of Canada (Med. Genetics) in 1994 and the Canadian College of Medical Geneticists (Clinical Genetics) in 1995.

His research activities in clinical chemistry have been focused in three areas. He has been part of the Familial Ovarian Cancer (FOC) Clinic team at the University Health Network since 1993 and active in its research programs. He was also co-investigator on a National Institutes of Health Familial Ovarian Tumor Study (1994-9). In this area he has been author or co-author of 17 related publications since 1993, including early papers identifying Canadian breast/ovarian cancer families linked to BRCA1/2 as well as keynote reviews of FOC syndromes, and new laboratory methods for BRCA1/2 mutation detection.

His second research interest has been the delineation of genetic and pathologic processes governing inherited bone and mineral disease. Among the conditions he has studied are: Morquio disease, inherited rickets related to altered Vitamin D receptors, primary hypomagnesemia, pseudohypoparathyroidism, and a new syndrome of osteogenesis imperfecta (OI) with hydrocephalus (Cole-Carpenter syndrome). His research has identified 3 hypercalcemic conditions (primary neonatal hyperparathyroidism, secondary hyperparathyroidism, and familial hypocalciuric hypocalcemia) that are part of a single allelic mutation, an Alu-insertion mutation in the calcium-sensing receptor (CASR) gene. He has also demonstrated that a common CASR polymorphism (Ala986Ser) is a principal determinant of circulating calcium concentrations in the general population, confirming clinical studies suggesting a strong genetic control of serum calcium levels. His third focus has been the biology of sulfur metabolism – more recently, hyperhomocysteinemia (hHcy) in vaso-occlusive disease. His lab found a very high rate of folate insufficiency and hHcy in cardiac transplant recipients (CTX), a group at high risk for an accelerated arteriosclerosis in the donor organ. His group has made the important observation that homocysteine levels in these patients depends not only on renal status and folate, but also correlates strongly with cyclosporine levels. He has published 159 abstracts, 90 peer reviewed full-length articles, 39 short reports. 22 editorials and solicited articles, and 27 book chapters.

Dr. Cole is a very deserving winner of this award and we wish him continued success in his very active research career into the future.