Dr. William Foulkes

Genetics reheats cold cases

The mystery began in 1976. Adolfo Pampena was diagnosed with a rare form of cancer, including a strange combination of symptoms and tumours in his stomach and colon. His perplexed medical team could not determine the cause of his disease or the risk for his descendents.

Thirty-five years later, the answers are outlined in a study led by investigators at the JGH Lady Davis Institute for Medical Research (LDI), the Research Institute of the McGill University Health Centre, and the McGill Program in Cancer Genetics at the Gerald Bronfman Centre for Clinical Research in Oncology. Senior author of the study—recently published in *The New England Journal of Medicine*—is Dr. William Foulkes, a genetics researcher at the LDI and the MUHC's Research Institute.

The researchers pinpointed a culprit gene, which is involved in regulating the separation of chromosomes. Instability during cell division can cause chromosomes to end up in the wrong place, leading to the development of tumours. "My father and family were relieved that the cancer risk for other relatives is much less than we thought," says Mary Pampena, Adolfo's daughter. "We now know what screening test to perform in the future."

In another study in the *Journal of the American Medical Association*, Dr. Foulkes describes solving a mystery involving five families with a long history of nontoxic multinodular goiter. Goiter, a thyroid disease, can lead to extreme swelling of the neck or larynx, but its cause is often iodine insufficiency and not genetics. While multinodular goiter was known to be genetic, no one had ever located the specific gene or mutation that was responsible—until now.

A team led by Dr. Foulkes and Dr. Marc Tischkowitz of the LDI and the Program in Cancer Genetics found that the mutation changes the protein in only one place. "A mutation in a disease gene usually causes many problems," says Dr. Foulkes, "but here we have no evidence it causes anything except goiter." He and his colleagues also confirmed an idea, first raised in 1974, that a genetic link exists between multinodular goiter and an unusual type of ovarian tumour.

Value of robotic surgery confirmed

Robot-assisted surgery, performed with technology such as the Jewish General Hospital's da Vinci Surgical System, dramatically improves outcomes in patients with uterine, endometrial, and cervical cancer, a JGH study has found.

With fewer post-operative complications and shorter hospital stays, robotic procedures also cost less, concludes the study, whose results were published in late 2010 in *The Journal of Robotic Surgery* and *The International Journal of Gynecological Cancer*.

"Patients' quality of life is dramatically improved," says Dr. Walter Gotlieb, who led the study and is Director of Gynecologic Oncology at the JGH Segal Cancer Centre. "They use far less narcotic pain medication—sometimes nothing stronger than Tylenol."

Robotic surgery technology was developed to overcome the limitations of minimally invasive surgery, including such notoriously difficult procedures as laparoscopy for cancer. "Laparoscopy is the gold standard of treatment for endometrial cancer," Dr. Gotlieb says, "but the learning curve is too steep for most surgeons. At the JGH, we went from only 15 per cent of our endometrial cancer patients undergoing laparoscopy to 95 per cent using robotic surgery. Previously, for cervical cancer, we did not perform minimally invasive surgery at all, whereas now all of our patients benefit from it."

Dr. Walter Gotlieb performs robot-assisted surgery.

In a letter to Dr. Joseph Portnoy, JGH Director of Professional Services, a patient with complex endometrial cancer, lavishly praised her "surgeon and his robot" and called the device a "marvel".

The da Vinci Surgical System was brought to the Jewish General Hospital in 2006 through the generosity and initiative of private donors.