## **TEACHING DOSSIER**

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## Associate Professor, Department of Pathology and Molecular Medicine, Queen=s University Director, DNA Diagnostic Laboratory Kingston General Hospital

## **1.0 Biographical Overview**

I currently hold two positions within the Queen's University Health Sciences Centre. I am Director of the DNA Diagnostic Laboratory at the Kingston General Hospital. My responsibilities include the reporting and supervision of molecular genetic testing, revision and writing of laboratory policies and procedures. Academically I hold the position of Associate Professor in the Department of Pathology and Molecular Medicine and accordingly there are expectations that I carry out research and participate in teaching and administration. I participate internally in administrative activities related to both positions, such as the hospital Radiation Safety Committee and the University Biohazards Committee. With respect to my clinical appointment I participate as either a member or chair of several government committees that oversee genetics services in this province or as a member of committees related to the training of genetics professionals. Both positions I hold provide opportunities to participate in broad array of educational activities for health care professionals, basic science students and the community.

## 2.0 Teaching Philosophy

## " The highest function of the teacher consists not so much in imparting knowledge as in stimulating the pupil in its love and pursuit" - Henri Amiel

The above quotation is one that I believe conveys the spirit of my approach to teaching. One aspect of teaching that I value greatly is the opportunity it gives me to get to know my students. I remember my student years and how much I appreciated not only receiving the lecturer's material but also in gaining insight into how they approached scientific problems and their views on life. I believe that a good teacher leads effectively, not by coercion or subjugation, but by example and by providing students with the tools to continue on their own. I would like students and fellows to obtain knowledge from me but also to understand why I conduct myself in the way that I do, and my approach to research and my service. I have had the best opportunity to practice this philosophy with those I have supervised in the laboratory as either basic science or clinical trainees because the contact with a student or fellow is prolonged over many months or years. Much of the course related teaching that I do for both basic science and medical students is limited to 10 hours or less per course, reducing the opportunity to get to know the students and leading to less satisfaction on my part. The concentrated teaching time necessitates that I transmit information efficiently, and importantly, encourage interest in the field of genetics. With respect to the medical students it will be many years before a student in first year medicine is in practice and the field of genetic diagnostics will change remarkably in that time. I would like medical students to remember genetics as an interesting topic. In the end I hope they remain receptive to advances in the field that will impact on their practice and to think of genetics as a possible cause when they encounter unusual cases, all of this hopefully to their patients' benefit.

## 3.0 Teaching Responsibilities

I am able through my academic and clinical appointments to participate in the teaching and training of both basic science and clinical trainees. I believe my practical experience with the clinical applications of molecular genetic technology, its successes, pitfalls and ethical considerations, allows me to bring a different perspective to the teaching of molecular genetics that is of value to any student regardless of their career path. For a summary of participation and dates for course related teaching please see Appendix 1.

## 3.1 Undergraduate Sciences Biology and Life Sciences Programs

My involvement in the undergraduate sciences program at Queen's has largely been as a supervisor of Biology 537 and Pathology 499 undergraduate thesis projects. The students I have supervised have worked on projects that have included the cloning of amyloid genes from Amphibia and Agnatha, analysis of the BRCA1 gene in familial breast and ovarian cancer, genetic studies of Type II diabetes, hemochromatosis, and the FMR-1 CGG repeat sequence in fragile X syndrome. I participate as a lecturer for Pathology 425, an undergraduate course in human genetics developed as a result of the increased demand for genetics in the undergraduate curriculum. For the last two years I have also participated in Biochemistry 441 Current Topics in Biochemistry a course that emphasizes discussion of science as well as associated ethical and social issues. Lastly, I participate in sessions that provide information on career choices in science to undergraduates when requested.

## 3.2 Faculty of Graduate Studies Program

## Pathology 825 - Human Genetics

I have both coordinated and taught in the Pathology 825 Human Genetics course. The primary objective of the course is to educate graduate students on a broad range of topics relevant

to the genetics of human disease. The material I have taught includes positional cloning, the human genome project, imprinting, molecular techniques, mutation analysis, pedigree analysis, risk analysis, population genetics and ethics.

#### Pathology 826 - Molecular Basis of Disease

The Pathology 826 Molecular Basis of Disease course seeks to educate students on the broad range of experimental studies that are pursued in the study of a given human disease. I have been involved in the presentation of the molecular basis of Prader-Willi and Angelman syndromes, the phenomena of genetic imprinting, and the molecular basis for the fragile X syndrome, Huntington's disease and Parkinson's disease.

#### Pathology 823 - Cancer Biology

The Cancer Biology course addresses the cellular, genetic and biochemical characteristics which underlie the development of cancer. I have been responsible for presenting material on the genetic basis of cancer, the mutator phenotype and genetic testing for cancer syndromes.

## 3.3 Faculty of Medicine

#### **Phase I Medicine:**

My involvement in this course has included curriculum development (discussed below), coordination, lecturing and the leading of small group sessions for the years 1992-2005. The lectures and small group sessions have included material on population genetics, molecular genetic technology, molecular diagnostics, familial cancer syndromes, risk analysis, and cytogenetics. I have been involved in the organization of several Medical Sciences Rounds on topics such as the fragile X syndrome, Prader-Willi syndrome and familial breast and ovarian cancer, hemochromatosis and forensic DNA analysis. I have also been responsible for coordinating the teaching of genetics in Phase I by members of the Division of Genetics.

In 2003 I was appointed as the Director of Phase I. My duties will be extended to coordinating the activities of the entire course. The duties include, working with course planners and coordinators, working with other Phase Directors and the Clinical Skills Director to ensure the integration of the course material, planning of examination content and participation in the evaluation of the students and the course, as well as attending Undergraduate Medical Education Committee meetings.

## Phase II Medicine:

For the upper years of the undergraduate medical school curriculum I have given or participated in lectures on genetics and ethics, cystic fibrosis, neurogenetics, thrombosis, hemochromatosis, and hemophilia. For the first time in 2003 I was a tutor for problem based learning in Phase IIA which requires a commitment of three hours per week for 14 weeks. I found the small group environment of PBL much more enjoyable and in keeping with the type of teaching role I like play.

## 3.4 Colleges and Institutes

## **Michener Institute:**

For the Michener Institute Post Diploma Program in Molecular Genetics I have for 12 years (1993-2005) acted as a supervisor of the clinical rotations of their trainees in the DNA Diagnostic Laboratory at the Kingston General Hospital. These rotations are of 6 or 12 weeks in duration and during that time a trainee is required to become familiar with the methodology and analysis of the genetic disorders for which our centre provides molecular genetic testing. I have been responsible for overseeing the training and the evaluation of those trainees. The time spent in discussions with the trainees about their laboratory studies and the relevant background material occupies approximately 2 hours per week of time.

## 3.5 Postgraduate Training

## **Canadian College of Medical Genetics:**

The Kingston General Hospital is accredited for the training of postdoctoral fellows for examination and certification by the Canadian College of Medical Genetics. My involvement at this centre is providing supervision of postgraduate fellows and training in human molecular genetics and molecular diagnostics (1996-2005).

## **Medical Residents:**

Each year I have been involved in the teaching sessions, for medical residents in hematology, pediatrics, geriatrics or pathology, specifically as an aid to their preparation for their specialty examinations. The teaching sessions are either one on one in the laboratory or given to a group.

## **3.6** Teaching in the Community

### High School Science Students and Teachers.

As part of course held at Queen's for High School Science Teachers I have given lectures on molecular genetics and its uses in medicine. I have also given tours and one on one training to individuals on request. As well I have participated in two enrichment courses for high school students offered at Queen's through Pathology and Cancer Biology. The material covered included molecular diagnostics and careers in genetics. **Senior Citizens:** 

In October 1999 I participated in a session attended by nearly 200 people on genetics and ethics as part of a lecture series on developments in science for senior citizens called ALater Life Learning≅, coordinated by Bunny Singer. Topics discussed included the human genome project, the patenting of human genes, and ethical issues in genetic testing.

#### Huntington Society of Canada

In November 2004, because of my role in the cloning of the gene for Huntington disease I was asked to give an overview of the current status of research on Huntington disease for the Kingston Chapter of the Huntington Society of Canada. This was done to help them restart the chapter's activities, which support local families affected by the condition.

## 4.0 Curriculum Development

#### 4.1 Michener Institute

Beginning in 1993 I became an advisor to a working group which developed the Michener Institute Post Diploma Program in Molecular Genetics which is only one of two programs which provide training for medical laboratory technologists performing molecular diagnostics in Canada.

#### 4.2 Phase I Medicine

In 1997 the first year medical school curriculum at Queen's University was restructured to fit into a 15 week term. I was part of the working group that designed the revised course outline and my most concentrated involvement was in the development of the first block of the Phase I curriculum, Cell, Genetics and Neoplasia.

Five years ago myself with Karen Harrison, Jennifer MacKenzie and Ines Sumargo applied to the Learning Faculty Associates for a project to develop a team approach to developing a new way to teach genetics to the undergraduate medical students. We did this because we had become increasingly concerned about how effective we were at communicating the genetics knowledge necessary for practising physicians. The project was approved and we began a collaboration with Elaine van Melle and Mark Fleming of the Learning Technology Unit. Within the condensed medical school curriculum at Queen's University, challenges include the volume of material, and a condensed timeframe. Discussions were initiated to evaluate the potential of using a web-based course program (Web CT) to organize and improve accessibility to course material and create an opportunity for students to apply these concepts in the evaluation and management of a clinical case. A great deal of time was spent on determining what our educational goals were and revising the objectives. Members of the team provided expertise in genetics, education and information technology. The team adopted the "ICE" Model (Ideas, Connections and Extensions), a theory that defines learning as a multistage process and encourages the development of higher order thinking skills. In addition to the use of the web site to provide access to reorganized course material, students were divided into small groups and required to review a case study posted on the site, and to email the instructors with a request for a genetic test and the rationale. Once requests were received, the test results were posted on Web CT. The groups were required to present an evaluation of their case and the implications of the genetic test results to their peers. We undertook an evaluation of the new curriculum and the results (Appendix 3) indicated that the students were receptive to the changes and were willing to participate. From our point of view as instructors we had a feeling of renewed enthusiasm for medical school teaching and a greater conviction that the material we were teaching them was applicable to medical practice. We have had several requests to present our teaching approach to others and have made several presentations locally, regionally to the Ontario Medical Education Network, and nationally at a Society for Teaching and Learning in Higher Education conference. The group received Honourable mention for the Alan Blizzard teaching award granted by the STHLE in 2002. The group also presented an abstract at the American Society of Human Genetics meeting in 2002.

I have had the opportunity as Director for Phase I to initiate further changes in the curriculum. Medical Science Rounds are a feature of the Phase I curriculum allowing the students an opportunity once per week to have a clinician and patient attend class to discuss the patient's condition and medical management. It had been unclear for sometime what the educational benefit of such sessions was and therefore discussion and revision was undertaken. The sessions were redesigned so the condition affecting the patient seen was directly relevant to the course material taught in lectures that were occurring just before or after. For half of the sessions we implemented small group follow up discussions of the pathophysiology that are led by pathologists and are held directly afterwards to reinforce what was just discussed with the patient and to encourage students in a directed way to acquire knowledge on their own. These sessions were helpful to their learning and encouraged their learning. Unsolicited comments from the students suggest they appreciated the format of the follow up sessions, the

manner in which the material was discussed by the pathologists made them feel like colleagues rather than students.

## 4.3 Curricular Renewal Task Force - School of Medicine

In June of 2004 a task force was struck to begin the process of redevelopment of the undergraduate medical curriculum. The work of revising the curriculum is in progress and the process includes a redevelopment of course objectives and an examination of effective methods of teaching which includes lectures, small group sessions, problem based learning and team based learning. The goal of the task force is to have a revised curriculum ready to be implemented in Fall of 2007.

## 5.0 Other Professional Activity Related to Teaching

## 5.1 Canadian College of Medical Genetics

I have served on the CCMG Examinations Committee for Molecular Genetics and this involved the setting, evaluation and administration of written and oral qualifying examinations to candidates (1996-2000).

## 5.2 Ontario Ministry of Health Laboratory Genetics Fellowship Selection Committee

As chair of the Ministry of Health Fellowship Selection Committee since 1996 I am responsible for coordinating the distribution, receipt and evaluation of the applications as well as for all communications with Ministry officials.

## 5.3 Ontario Advisory Committee on Genetics

The Ontario Ministry of Health and Long Term Care has established a new provincial committee to advise the government on policy related to the genetics practise in the province. I was asked to be on this committee because of my molecular genetics expertise and my experiences with setting up testing programs in the province. Recently I have also joined the Education Subcommittee. This committee has been asked to develop goals for the education health care providers, medical and nursing students and the public in the clinical applications of genetics. We have been asked to begin to pursue what is thought to be the most realizable of the goals and that is to develop genetics continuing medical educational materials for health care providers that can be distributed provincially. This work led directly to the establishing of the Genetics Education Project described in the next section. This group will also be called upon by the MOHLTC to evaluate genetics related educational materials developed by other branches of the Ministry.

## 5.4 The Genetics Education Project

In 2004 the OACG Education Subcommittee received \$502,000 in funding from the Ontario Women's Health Council to implement the Genetics Education Project. The primary objectives of the project are to facilitate the delivery of genetics services by primary care providers and to develop effective models for genetics education. A secondary objective is to facilitate informed decision-making for women contemplating prenatal genetic testing. Central to our proposal is a commitment to apply appropriate ethical principles to all educational models and materials related to these initiatives. Important considerations included the need to respect the beliefs, cultural traditions, circumstances and feelings of each individual; to provide clear information (including alternatives and consequences) that enables informed, independent, coercion-free decision-making; to refer to appropriate specialists when indicated; to maintain client confidentiality; and, to avoid exploiting patients. The target group for the project is the primary care providers of Ontario, including family physicians/general practitioners, nurses and midwives, and other specialists (including pediatricians, internists, obstetricians/gynecologists and general surgeons) who provide primary care. The secondary target is the public of Ontario.

To date we have produced learning modules for the Foundation for Medical Practice Education on hemochromatosis and prenatal screening tests. We are in the process of developing brochures for providers and the public on prenatal screening tests for Down's Syndrome (Appendix 4) and have developed audiovisual materials for Alzheimer disease, hemochromatosis, colon cancer, breast cancer and prenatal screening.

In November 2005 we will be conducting a Peer Presenter Workshop in Toronto. The goal of the workshop is to educate a small number of influential health care providers throughout the province about genetics so that they can return to their communities and teach about this area. Attending this program are individuals interested in genetics, identified by each of the Ontario Medical Schools, from the disciplines of family medicine. One of the activities will be the use and presentation of the audiovisual materials that have been developed by the group.

## 6.0 Publications and Presentations

## 6.1 Abstracts

Taylor, S., MacKenzie, J., Harrison, K., Sumargo, I, Fleming, M., VanMelle E. Enhancing genetics education in medical school; An approach using a web-based course program. Annual meeting American Society of Human Genetics 2002, Abstract #1019.

VanMelle, E., Taylor, S., MacKenzie, J.

Using a team approach to enhance student learning through the use of technology in a first year

medical genetics course. Society for Teaching and Learning in Higher Education Annual Meeting, 2002, Abstract #149.

## 6.2 Invited Presentations

Ontario Medical Education Network (OMEN) Education Grand Rounds April 24, 2003

"Enhancing genetics education in medical school: An approach using a Web-based course program"

Eastern Ontario Society for Education and Technology December 2, 2003

"Enhancing genetics education in medical school: An approach using a Web-based course program"

## 7.0 Evidence of Teaching Effectiveness

## 7.1 Self Assessment

I like teaching and have liked the individuals I have taught. I am also interested in the material that I teach. I believe this comes across in my teaching, I think it would be rare to find someone who said I wasn't enthusiastic. Where I do less well is in my presentation style. I either speak too quickly or misjudge the background of those I am teaching, both of which can leave students confused, hence I always make it clear to my students that I am available to answer their concerns outside the class. Presentation style is something I constantly have to work at but believe that I am getting better at it as time passes. If I am going to teach I also believe in being prepared and if I have any regrets about my teaching efforts they usually stem from conflicts with my other responsibilities that reduce the time available to me for preparation. I have tried to update and improve the material I teach each year, streamlining as much as possible. In terms of professional development related to teaching in February 2000 I attended a WebCT designers course and workshop on writing multiple choice questions given by the Faculty of Medicine in October 2002.

## 7.2 Assessments by Others

Appended to this dossier is documentation of my teaching abilities by others as well as relevant correspondence (Appendix 5).

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# **Appendix 1. Summary of participation in undergraduate and graduate courses**

Table 1. Summary of participation in courses taught at Queen's University, C= coordinator, D=Director, L= lecturer, T=tutor.

Course	# of students	Years taught	Objectives/Topic
Undergraduate Basic Science			
1) Pathology 499 Thesis supervision	1	1993-1994 1996-1997 2001-2002 2003-2004	To provide practical laboratory experience to basic science undergraduates
2) Biology 537 Thesis supervision	1	1992-1993 1995-1996 1997-1998 1999-2000 2001-2002 2002-2003	As above

		2005-2006	
3) Pathology 425 (L)	15	2003,2004 2005	To provide exposure to basic human genetics and current relevant topics in the field.
4) Biochemistry 441	15	2004, 2005	Clinical applications of molecular genetics
Undergraduate Medicine			
1) Phase I (L, C) (D)	75-100	1992- present 2003- present	<ul> <li>-Human genetics, molecular genetics, cancer genetics</li> <li>- curriculum planning and coordination</li> </ul>
2) Phase IIA (L)	75	1998, 1999	-Inherited disorders of hemostasis
3) Phase IIA (T)	75-100	2003	-problem based learning in medicne
4) Phase IIB (L)	75-100	1993, 1995, 2000-present	-Inherited neurological disorders, genetics of trinucleotide repeat disorders
Course	# of students	Years taught	
			Objectives/Topic
Undergraduate Medicine cont'd			
5) Phase IIC (L)	75	1993, 1994	- genetics of cystic fibrosis
6) Phase IIE (L)	90	2001	- congenital disorders

7) Medicine in Society	75-100	1994, 1997, 1998	- genetics and ethics
(L) Graduate			
<b>Basic Science</b>			
1) Pathology 823 Cancer Biology (L)	10-15	1998, 2000, 2002, 2004	Relevant topics in cancer biology, genetics, epidemiology and pathology
2) Pathology 825 Human Genetics (C, 1994, L)	10-15	1994, 1996, 1998, 2000, 2002, 2004	Genetics of human disease, including relevant topics such as the human genome project
3) Pathology 826 Molecular Basis of Disease (L)	10-15	1997, 1999, 2001, 2003 2005	Indepth discussion of approaches used to study human disease

## **Appendix 2: Summary of Supervisory and Examination Activities**

#### **Postdoctoral Fellows:**

Dr. Yagang Xie,	1996-1998
	Currently, Director of the Molecular Genetics Laboratory of the
	Memorial University Health Sciences Centre, Newfoundland
Dr. Harriet Feilotter	,1998-2000
	Current, Associate Director, DNA Diagnostic Laboratory Kingston General Hospital and Director, Microarray Facility, Queen's University
Dr. Alain Lagarde,	1998-2000 Laboratory Scientist, Ottawa General Hospital
Dr. Shulin Zhang,	2004-2007 Current Canadian College of Medical Genetics Fellow Recipient of a Ministry of Health Laboratory Genetics Fellowship

## Graduate Studies:

## **Supervisory Committee:**

#### Past:

Lara Boccia, M.Sc., Pathology Emily Croteau, M.Sc., Biology Angela Keightly, Ph.D., Pathology Sarah Kinkley M. Sc., Pathology Brian Pak, Ph.D, Anatomy and Cell Biology Anona Patriana, M.Sc., Biology Andrea Smith, Ph.D., Biology Mary Stapleton PhD, Biology Jennifer Struthers, M.Sc., Pathology Mary Vallianatos, M.Sc., Biology Brian Weese **Current:** Brian Weese, Biology

#### **Examinations Committee:**

Nazareth Bastajian, M.Sc, Biochemistry Andrea Davidson, Ph.D., Biology Sarah Ely, M.Sc. Pathology Lee Fraser, M.Sc., Pathology Carmen Gervais, MiniMasters, Biology Carmen Gervais, Ph.D., Biology Jennifer Glegg, M. Sc, Biology Taranjit Gujral, M.Sc., Pathology Martin Kang, M.Sc, Pathology Angela Keightly, MiniMasters, Pathology Alexander Langstratz, Ph.D, Pharmacology Leah Lazaruk, M.Sc., Pathology Eva Lin, MSc, Pathology Jennifer McVeigh, M.Sc., Pathology Julie Shaw, M.Sc, Pathology Peter Truesdell, Ph.D. Comprehensive, Pathology JoAnna Walker, M.Sc., Pathology Cathy Watson, Ph.D., Biochemistry Barry Weese, M.Sc, Biology Susan Wood, Phh.D, Biology Jiping Zhao, M.Sc., Biology

#### **Undergraduate Sciences Program:**

#### **Thesis Supervision:**

Julia Davidson-Arnott, Biology Julia Frei, Biology Emma Goodall, Biology Germaine Hao, Pathology Hannah Hoag, Pathology Sarah Kinkley, Biology Evan Merrick, Biology Stephanie Vanderpol, Pathology Simmy Wan, Pathology Shannon Wesley, Biology Elisabeth Whiting, Biology Paula Williams, Biology

### **College Programs Practical Rotation Supervision:**

#### **Michener Institute:**

Krista Croshuk Andrew Deck Nicole Fabricus Pat Florio Melissa Kell Sean Kim Kelly Maropakis Donna Maybury Sylvanna Moser Ray Okamoto Karen Persad Marlene Sharma Alan Stewart Lynda Walker Wendy McCaul **Daimion Fumerton** 

## **Appendix 3: The Medical Genetics Learning Technology Team Process**

## CONTEXT

The changes in the Medical school curriculum which saw a decrease in the mumber of teaching hours available and the explosion of information on the genetic basis of disease created a conflict in terms of what to teach medical students. The genetics curriculum for a time became focused on presenting students with as much information as possible in as short a time as possible, leading to frustration for both the students and the faculty. The faculty, all of whom have clinical appointments, recognized the increasing relevance of genetics to family physicians and thus the need to reorganize the curriculum to make it more clinically relevant. What the group did, that is apparently still a unique phenomena in medical education was to work with experts in education to identify education objectives. To consciously make the decision to not teach everything but to tailor the content to the genetics knowledge and skills that would be necessary for a practicing physician. What was also unique at the time was the combining of the didactic and web based portions of the course and to use the technology to facilitate and increase interaction between the instructors and the students rather than merely as a means of reducing faculty workload. The group spent a year planning and implementing the changes in the genetic curriculum. An outline of the planning, implementation and evaluation of the curriculum changes is given below. As part of the evaluation we felt it was important to determine if we had met our objective of making the students more aware of the process of genetic assessment and management.

## **REFLECTION AND PLANNING**

What learning outcomes did we expect?

Acknowledged that we couldn't turn all medical students into geneticists.

<u>But</u> we could give them some basic tools and an increased understanding of how genetics is used in clinical practice. The objectives for the course were revised to reflect this change in approach. Ultimately we wanted the medical students to,

- Have an appreciation of the role of genetics in human disease
- Appreciate the concept of genetic risk
- Be more likely to recognize the presence of a genetic condition in an individual or family
- Increase their confidence in the use of genetics

#### HOW WERE THE CURRICULUM CHANGES ACHIEVED Implementation

The team adopted the "ICE" Model (Ideas, Connections and Extensions), a theory that defines learning as a multistage process and encourages the development of higher order thinking skills. The course material was reorganized to have basic facts presented in didactic lectures, and the concepts of genetic assessment and management addressed through the use of clinical cases that were accessed using WEBCT and discussed in small group sessions. The students were meant to receive information in the class that would help them assess their cases while they worked on them. The students were given their cases to work on in small groups. They were required to request an appropriate genetic test for their case, via email and were sent the laboratory results and reports upon receipt of the request. They then had to present their case to their peers, who had been assigned different cases, as part of a case rounds. What we hoped to duplicate was that the approach the students would have to take and the issues they

would have to consider would be close to what they would be in everyday clinical practice.

<u>I</u> deas	$\rightarrow$	<u>C</u> onnec	tions	$\rightarrow$	<u>Extensions</u>
basic facts	rel	lationsh	ips		application
Web CT	$\rightarrow$	$\rightarrow$	$\rightarrow$		Web CT and Email
Organize in	formation			Analy	sis of clinical cases
Timely acco	ess			Posti	ng test requests and case summaries
Exposure to	o other webs	ites			

## **GENETICS CASES DEVELOPED**

Cases which differed in their genetic etiology were developed, accompanying the cases were different psychosocial issues that also needed to be addressed which are typical of what is encountered clinically.

Numerical and Translocation Down's syndrome (trisomsy 21)

- different reproductive issues, counseling issues

Translocation Trisomy 13q

- prenatal testing, parental options and counselling.

Fragile X Syndrome (unstable trinucleotide repeat) - dysfunctional family

Prader-Willi Syndrome (chromosome 15 imprinting) - parental denial

Gaucher Disease (autosomal recessive, metabolic disorder)

- prenatal diagnosis, possible at risk pregnancy

Marfan's Syndrome (autosomal dominant, connective tissue disorder)

- problems with evaluation, key individuals deceased.

Breast cancer (familial cancer susceptibility)

- use of provincial guidelines

## TRISOMY 21 CASE, CYTOGENETIC RESULTS AND REPORT INCLUDED

#### **EVALUATION**

Were objectives met, how were curriculum changes received?

## 1) Class Participation/Compliance

Test requests submitted	100% of groups (2001-2005)		
Summaries posted	2001 67% 2002 40%		

The decrease in the number of case summaries posted was felt to be the result of the increased workload associated with a new MSR format. This indicated to the group that when implementing new exercises they have to be done in the context of the other workload in the course. The overall decrease in satisfaction with the genetics portion of the curriculum, given below, was also taken as evidence of this.

## 2) Evaluation - Test requests

## Example of content from test requests for Down Syndrome Case - Trisomy 21

## Poorer quality of test request

"Our group has decided that for our case the patient has symptoms of Down syndrome. We suggest a genetics test for trisomy 21.

## Higher quality test request

Paraphrased:

"Request a karyotype for trisomy 21, because of the following clinical features.."

"disorder does not show Mendelian inheritance, incidence of Downs 1/300 according to maternal age"

"Fish test for chromosome 16p deletion if karyotype normal, to look for Rubenstein-Taybi syndrome, because of the following clinical features.."

"Inform parents of strong family history of breast cancer, future testing for mutations of BRCA1 and BRCA2 genes."

The test requests were given a score out of 5 for inclusion of an evaluation of the diagnosis, the rationale for requesting a particular test and possible outcome and justification for their choice. Evaluation of test requests submitted by students

## Test requests (Maximum score = 5) diagnosis, rationale, test, possible outcome

	2001	2002-2004
#sections/year (5groups/section)	3	5
Fragile X	3.7	4.5
Translocation Trisomy 21	4.7	4.2
Breast Cancer		4.3
Prader- Willi Syndrome		3.9

Comment: most often students had trouble with the actual expected outcome of the test, lack of background in genetics.

Group vs Individual performance

in 2004 students were given the choice of working in a groups or alone, 17 students of the 100 chose to work individually.

	#requests	Range	Mean± S.D.
Groups	13	2.5-5.0	$4.3 \pm 1.01$
Individual	17	3.0-5.0	$4.7 \pm 1.29$

When given a choice working in a group was preferred, in 2005 all students chose to work as a group.

## 3) Evaluation - Case Summaries

The posted case summaries in 2001 were evaluated as to whether or not the implications for the patient and their family were identified and an appropriate follow up suggested. The results below were obtained, they would suggest that the majority of groups identified the issues that were appropriate for their patients and families and were able to suggest a management strategy. The only issues not addressed adequately were the reproductive issues for the patients, possibly the result of the instructors omitting this from the discussion of clinical issues in the didactic portion of the course.

Implications for patient	prognosis	17/18
	reproductive	7/14
	psychosocial	15/18
Implications for family	carrier risk	17/18
members	reproductive	13/14
memoers	psychosocial	18/18
Treatment/followup	treatment	16/18

## 4) Formal Evaluation Results - Questionnaire given to students, summary of results and comments attached.

Year (Response Rate)	<u>2001</u> (64%)	<u>2002</u> (58%)
Genetics background	70.3%	79.3%
Ability to follow material, easy	63.8%	55.2%

Used Web CT to access material	100%	91%
Active participation in group discussions	93%	70.6%
Overall satisfaction	91.3%	69%

## Anecdotal Results / Unintended Outcomes

-Students better prepared for class

-Higher quality of questions

-Increased faculty satisfaction with teaching

-Renewed commitment to teaching

-Sharing of model with other faculty members

-Influence of other course changes - negative

## Benefits of the LTT approach

-Usefulness of combined expertise in pedagogy technology and in the subject to be taught -Implementation of technology resulting in a better quality of instructor/student interaction -No increase in teaching hours, little workload to maintain.

# APPENDIX 4:DRAFT OF BROCHURE FOR HEALTH CAREPROVIDERS ON PRENATAL TESTING FOR PROVIDERS

The attached brochure is a draft of a brochure being developed to educate health care providers on the availability of prenatal testing procedures in the province of Ontario.

## **Appendix 5: Evaluation of Teaching Effectiveness**

The attached teaching and evaluations are organized as follows:

1. Evaluations

Phase I WEBCT Based Instructor Evaluation by Undergraduate Office Evaluations by Genetics Group 2001 and 2002
Phase IIA Problem Based Learning Evaluation by Undergraduate Office – 2003
Pathology 823 – Cancer Biology 2004
Pathology 430/826 – 2005
Resident Teaching Evaluation

2. Correspondence related to teaching

Letter from Dr. C. Padfield Email from Pathology Graduate Student Fiona Rawle Letter from Isla Horvath, Huntington Society of Canada