



McGILL UNIVERSITY
Faculty of Medicine
School of Communication Sciences & Disorders
Winter 2020

SCSD 688
Genetics in the Practice of Speech-Language Pathology
1 Credit
Online work: Self-directed learning, January 6 – 21
Tutorial: Tuesday, January 21, 2:15 – 3:15 pm, Room 869
Class Times: Tuesdays, January 28 and February 4, 11 and 18, 1:45 – 4:30 pm
Location: SCSD, Rooms 862 and 869

Instructor: Jennifer Fitzpatrick, MS, CGC
Assistant Professor, Department of Human Genetics
Office: Strathcona Anatomy and Dentistry, Room W-315C; **Office Phone:** 514-398-4400 ext 00704
Office Hours: By appointment; e-mail is also welcomed
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COURSE CONTENT:

Normal human genetics and embryological development, their relationship to congenital disorders that affect communication, and implications for speech-language pathology practice.

LEARNING OUTCOMES:

ROLE 1: CENTRAL ROLE/CONTENT EXPERT

1.1 Foundational principles	
a. Apply basic knowledge of biomedical, cognitive, linguistic, pharmaceutical, physical and socio-behavioural sciences relevant to human communication processes, including knowledge of their acoustic, biological, cultural/linguistic, developmental and neurological bases.	
Demonstrate a basic understanding of normal human genetics and embryological development, and their relationship to congenital disorders that affect communication.	Online learning module, tutorial
Describe the features of Down syndrome and identify those that impact speech and language.	Lecture
Understand the importance of family history (minimum three generations) in assessing predisposition to disease.	Role plays
Distinguish the various laboratory methods used in genetic testing.	Lecture, role plays
d. Apply knowledge of hearing, hearing loss and disorders of the auditory system, relevant to practice as a speech-language pathologist.	
Recognize common single gene causes of hearing loss.	Lecture, role plays

1.3 Assessment	
a. Develop an assessment strategy to evaluate communication.	
Gather genetic family history information, including an appropriate three-generation pedigree.	Role plays
Recognize the signs of a potential underlying genetic condition.	Lecture, role plays

ROLE 2: COMMUNICATOR

2.1 Oral and written communication	
c. Provide relevant information.	
Provide appropriate information about the potential risks, benefits, and limitations of genetic testing.	Role plays
Explain effectively the reasons for and benefits of genetic services.	Role plays
d. Listen actively.	
Address the psychosocial implications of a diagnosis or potential diagnosis of a genetic condition in a family	Role plays

ROLE 4: ADVOCATE

4.1 Client advocacy	
b. Advocate for individual clients where appropriate.	
Identify and refer clients who might benefit from genetic services.	Role plays

Roles 3, 5, 6 and 7 are not specifically addressed in this course.

INSTRUCTIONAL METHOD:

In this course, students will review basic principles of human genetics through online materials selected for their appropriateness for speech-language pathologists. After successful completion of the online module, students will participate in a series of learning activities designed to apply these concepts as well as relevant aspects of embryological development to clinically relevant case scenarios. Over 4 in-class half-days, students will attend a series of short didactic presentations, alternating with learning activities and discussion. Learning activities will include role plays and simulations to develop skills in pedigree taking, addressing with clients the potential for an underlying genetic condition, describing the benefits and limitations of genetic assessment and testing, and referring clients to genetics professionals. Additional time will be devoted to developing beginning-level listening skills to address the response of clients to the possible or known genetic diagnosis in a family such as guilt, shame, anger, sadness, denial and/or frustration over the ambiguity of the undiagnosed child.

COURSE MATERIALS:

Required readings will be available through MyCourses. A combination of free online resources and textbook chapters available through the McGill Library will be linked to the course site.

COURSE SCHEDULE:

Date	Self-Directed Learning	Instructor
Jan 6 - 21	Online work - Self-directed learning on basic mechanisms of genetics and inheritance patterns Completion of readings and problem set in MyCourses	N/A
Jan 21 2:15 – 3:15	Tutorial to address any areas of difficulty in online readings and problem set	J. Fitzpatrick
Jan 21 4:00 – Jan 27 12:00 pm	Quiz open in MyCourses	N/A
Jan 28	In-Class Session 1	Instructor
1:45 – 2:15	Introduction to course The range of influence of genetics in medicine - Chromosome abnormalities	J. Fitzpatrick
2:15 – 2:45	Case Study: Trisomy 21, Down syndrome	L. Russell
2:45 – 3:00	Clinical laboratory tests for chromosome abnormalities	J. Fitzpatrick
3:00 – 3:15	Break	
3:15 – 3:30	The medical genetics assessment	J. Fitzpatrick
3:30 – 3:45	Discussion - Challenges of clinical training, the struggle to perform perfectly	J. Fitzpatrick
3:45 – 4:15	Role play exercises - Addressing parental reactions to a genetic diagnosis such as denial, sadness, anger, guilt or shame	L. Baret J. Fitzpatrick
4:15 – 4:30	Quiz	
Feb 4	In-Class Session 2	
1:45 – 2:15	The range of influence of genetics in medicine – Single gene inheritance	J. Fitzpatrick
2:15 – 2:45	Craniofacial disorders	L. Russell
2:45 – 3:00	Break	
3:00 – 3:10	Clinical laboratory tests for single gene disorders	J. Fitzpatrick

3:10 – 3:30	Hereditary hearing loss	L. Whelton
3:30 – 4:15	Role play exercises and case study: Gathering family history information, constructing pedigrees	J. Fitzpatrick L. Whelton
4:15 – 4:30	Quiz	
Feb 11	In-Class Session 3	
1:45 – 2:10	The range of influence of genetics in medicine – Chromosome microdeletion syndromes Clinical laboratory tests for chromosome microdeletion syndromes	J. Fitzpatrick
2:10 – 2:30	Del 22q11 – DiGeorge syndrome	L. Whelton
2:30 – 2:40	Further genetics	J. Fitzpatrick
2:40 – 2:55	Break	
2:55 – 3:00	Review of selected quiz 3 questions (if necessary)	J. Fitzpatrick
3:00 – 3:15	Case study pedigree exercise: Baby Maria's family	J. Fitzpatrick
3:15 – 4:15	Case study: Baby Maria's family Pedigree exercise Role plays - Assess the potential for a genetic diagnosis in a family, refer a client to genetic services	J. Fitzpatrick L. Whelton
4:15 – 4:30	Quiz	
Feb 18	In-Class Session 4	
1:45 – 2:15	The range of influence of genetics in medicine - Multifactorial inheritance, de novo mutations and epigenetics Laboratory detection of genetic changes for complex traits	J. Fitzpatrick
2:15 – 2:45	Autism	J. Fitzpatrick
2:45 – 3:00	Break	
3:00 – 3:30	Informed consent in genetic testing, including genetic variants of uncertain significance	J. Fitzpatrick
3:30 – 4:15	Role play exercises - Addressing ambiguity and the undiagnosed child, explaining the genetic testing process	J. Fitzpatrick
4:15 – 4:30	Quiz	
Deadline Mar 10	One-hour final assessment, to be completed in MyCourses	

EVALUATION:

20%	Mastery of pre-course online content as evaluated by an online quiz with a grade of at least 80%. Students may take the quiz multiple times.
40%	Quizzes after each in-class session (4, each valued at 10%)
40%	Final assessment – online test with multiple choice and short-answer questions

McGILL POLICY STATEMENTS:

Academic Integrity

McGill University values academic integrity. All students must understand the meaning and consequences of cheating, plagiarism and other academic offences under the Code of Student Conduct and Disciplinary Procedures (see www.mcgill.ca/students/srr/honest/ for more information).

Language of Submission

In accord with McGill University's Charter of Students' Rights, students in this course have the right to submit in English or in French any written work that is to be graded.

Protection of Course Materials

Instructor-generated course materials (e.g., handouts, notes, summaries, exam questions, etc.) are protected by law and may not be copied or distributed in any form or in any medium without explicit permission of the instructor. Note that infringements of copyright can be subject to follow up by the University under the Code of Student Conduct and Disciplinary Procedures.

Inclusive Learning Environment

As the instructor of this course I endeavor to provide an inclusive learning environment. If you experience barriers to learning in this course, do not hesitate to discuss them with me and the [Office for Students with Disabilities](#), 514-398-6009.

End-of-Course Evaluations

[End-of-course evaluations](#) are one of the ways that McGill works towards maintaining and improving the quality of courses and the student's learning experience. You will be notified by e-mail when the evaluations are available. Please note that a minimum number of responses must be received for results to be available to students.

Preparedness for a Potential Pandemic

In the event of extraordinary circumstances beyond the University's control, the content and/or evaluation scheme in this course is subject to change.