



Fall 2023

SCSD 688
Genetics in the Practice of Speech-Language Pathology
1 Credit
Online work: Self-directed learning, Aug 30 – Sep 29
Tutorial: Friday, Sep 29, 1:15 – 2:15 pm
Class Times: Fridays, Oct 13 & 27, Nov 3 & 10, 1:15 – 4:00 pm
Location: SCSD, Rooms 862 and 869

Instructor: Jennifer Fitzpatrick, MS, CGC
Assistant Professor, Department of Human Genetics
Office: Strathcona Anatomy and Dentistry, Room 2/38C; Office Phone: 514-398-1649
Contact: Phone calls and e-mails are welcomed. Meetings can be arranged upon request.
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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:

1: ROLE OF EXPERT

1.1 Knowledge Expert	
b. Apply basic knowledge from relevant fields that apply to communication and feeding and swallowing across the lifespan.	
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
c. Apply knowledge of hearing, hearing loss and disorders of the auditory system to the practice of speech-language pathology.	

Recognize common single gene causes of hearing loss.	Lecture, role plays
1.2 Clinical Expert	
f. Plan, conduct and adjust an assessment	
Gather genetic family history information, including an appropriate three-generation pedigree.	Role plays
g. Analyze and interpret assessment results.	
Recognize the signs of a potential underlying genetic condition.	Lecture, role plays
h. Develop and share recommendations based on assessment results.	
Identify and refer clients who might benefit from genetic services.	Role plays
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

2: ROLE OF COMMUNICATOR

a. Communicate respectfully and effectively using appropriate modalities.	
Address the psychosocial implications of a diagnosis or potential diagnosis of a genetic condition in a family	Role plays

3: ROLE OF COLLABORATOR

a. Establish and maintain effective collaborations to optimize client outcomes.	
Identify and refer clients who might benefit from genetic services.	Role plays

Roles 4, 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
Aug 30 – Sep 29	Online work - Self-directed learning on basic mechanisms of genetics and inheritance patterns Completion of readings and practice problem set in MyCourses	N/A
Sep 29 1:15 – 2:15	Tutorial to address any areas of difficulty in online readings and problem set	J. Fitzpatrick
Sep 29 4:00 pm - Oct 13 2:30 pm	Quiz open in MyCourses	N/A
Oct 13	In-Class Session 1	Instructor
1:15 – 1:45	Introduction to course The range of influence of genetics in medicine - Chromosome abnormalities	J. Fitzpatrick
1:45 – 2:15	Case Study: Trisomy 21, Down syndrome	Y. D’Souza
2:15 – 2:30	Clinical laboratory tests for chromosome abnormalities	J. Fitzpatrick
2:30 – 2:45	Break	
2:45 – 3:00	The medical genetics assessment	J. Fitzpatrick
3:00 – 3:15	Discussion - Challenges of clinical training, the struggle to perform perfectly	J. Fitzpatrick
3:15 – 3:45	Role play exercises - Addressing parental reactions to a genetic diagnosis such as denial, sadness, anger, guilt or shame	J. Fitzpatrick
3:45 – 4:00	Quiz	
Oct 27	In-Class Session 2	
1:15 – 1:45	The range of influence of genetics in medicine – Single gene inheritance	J. Fitzpatrick
1:45 – 2:15	Craniofacial disorders	L. Russell
2:15 – 2:30	Break	
2:30 – 2:40	Clinical laboratory tests for single gene disorders	J. Fitzpatrick

2:40 – 3:00	Hereditary hearing loss	J. Fitzpatrick
3:00 – 3:45	Role play exercises and case study: Gathering family history information, constructing pedigrees	L. Baret J. Fitzpatrick
3:45 – 4:00	Quiz	
Nov 3	In-Class Session 3	
1:15 – 1:40	The range of influence of genetics in medicine – Chromosome microdeletion syndromes Clinical laboratory tests for chromosome microdeletion syndromes	J. Fitzpatrick
1:40 – 2:00	Del 22q11 – DiGeorge syndrome	L. Whelton
2:00 – 2:10	Further genetics	J. Fitzpatrick
2:10 – 2:25	Break	
2:25 – 2:30	Review of selected quiz questions (if necessary)	J. Fitzpatrick
2:30 – 2:45	Genetic red flags	J. Fitzpatrick
2:45 – 3:45	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
3:45 – 4:00	Quiz	
Nov 10	In-Class Session 4	
1:15 – 1:45	The range of influence of genetics in medicine - Multifactorial inheritance, epigenetics Laboratory detection of genetic changes for complex traits	J. Fitzpatrick
1:45 – 2:15	Autism spectrum disorder	J. Fitzpatrick
2:15 – 2:30	Break	
2:30 – 3:00	Informed consent in genetic testing, including genetic variants of uncertain significance	J. Fitzpatrick
3:00 – 3:45	Role play exercises - Addressing ambiguity and the undiagnosed child, explaining the genetic testing process	J. Fitzpatrick
3:45 – 4:00	Quiz	

EVALUATION:

- 20% Mastery of pre-course online content as evaluated by an online quiz with a minimum grade of 80%. Students may take the quiz multiple times.
- 40% Quizzes during in-class sessions (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.

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.

Instructor-Generated Course Materials and Copyright

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