

THE DIVISION OF MEDICAL GENETICS

Royal University Hospital, Saskatoon, Saskatchewan

Adult Genetic Services

Genetic counselling for adult onset disorders is an area that has expanded significantly in recent years. With the advent of new DNA diagnostic techniques and the discovery of disease genes, more families are interested in establishing a diagnosis and learning about their risk of developing diseases that have been in the family for generations. Genetic counselling can help diminish a patient's anxiety and remove uncertainty. Genetic counselling prior to testing allows individuals the chance to make informed decisions about medical treatment (including genetic testing) and reproductive choices.

It is important to recognize that the expression of certain genetic conditions can vary widely between individuals and families, and among members of the same family. Sometimes DNA tests may be the only way to establish whether or not someone is or will be affected.

Genetic testing in an individual who is currently asymptomatic for a disorder is called predictive or presymptomatic testing. We follow a strict protocol for presymptomatic testing which includes having the patient attend two separate appointments. We highly recommend that patients attend the appointments with a trusted support person, preferably someone who is not a blood relative. The purpose of the first appointment is to review the family history and personal medical history in order to discuss the risks, benefits, and choices regarding testing in an individual manner. If the person chooses to have the testing, a second appointment is arranged for disclosure if and when the patient chooses to receive them. Results will only be given in person.

It has been well documented that patients have strong emotional reactions to both positive and negative results. The Genetic Counsellor will help clarify the implications of the results, discuss future screening or treatments available and how other family members may benefit from genetic counselling and/or testing.

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