

THE DIVISION OF MEDICAL GENETICS

Royal University Hospital, Saskatoon, Saskatchewan

FAQs FOR PATIENTS

Q. When do people go to see Medical Genetics?

- A. Patients are often referred when major changes are taking place in the family structure. Some examples include:
- When an individual who is affected with or is a carrier for a genetic or chromosomal condition reaches reproductive age
 - When a pregnancy has been identified by ultrasound to have abnormalities or the mother will be 40 years of age or older at the time of delivery (younger if there are multiples!)
 - When a child is born with a birth defect or serious medical condition
 - When significant developmental delays or behavioural concerns arise
 - When other family members are planning to have children and a definitive diagnosis is needed for appropriate risk counselling to be provided
 - When an individual is diagnosed clinically with a possible or definite genetic disorder for which testing is now available

Q. What is genetic counselling?

- A. Genetic Counsellors and Medical Geneticists work as part of the healthcare system. Their job is to provide you with information and support when a member of your family has been diagnosed, either in our clinic or elsewhere, with a condition that may have a genetic component. Genetic professionals will take the time to explain the diagnosis to you. They will let you know if other family members are at risk. Tests to confirm the diagnosis may be arranged. Genetics professionals are able to provide insight into some of the difficulties that families may have following diagnosis of a genetic condition. They can review available surveillance and treatment options with you and make referrals to other medical specialists that can help care for you and your family.

Q. Where are clinics held?

- A. Clinics are held in Saskatoon at the Royal University Hospital. If you are from out of province, please visit the Canadian Association of Genetic Counsellors website at www.cagc-accg.ca to find the genetics center closest to you or ask your doctor for a referral.

Q. What do I need to do to prepare for a genetics appointment?

- A. To prepare for a medical genetics appointment you will be sent a family history form to complete. In order to do this, you will need to gather as much information as possible about your family history. Please return the completed forms to Medical Genetics. An appointment will be booked once the family history forms are returned. Many personal traits are inherited. These traits include hair and eye colour, height and intelligence, as well as risks for certain genetic conditions and health problems. It is important for individuals to learn as much about their medical family history as possible. By looking at your family history we can help to predict which health problems you may be at increased risk for in the future and also may be able to help you to reduce some of these risks.

Q. How do I go about obtaining my family history?

- A. Good places to look for information on your family history include your relatives (parents and older relatives are often good sources of information), baby books, birthdate books, and family bibles. There are now many websites dedicated to putting together family trees. Some family members may be more reluctant than others to share their personal medical history. Be respectful of this. Everyone deals with their health in different ways.

Q. What about my privacy?

- A. The information that you provide will be stored in a chart in the Division of Medical Genetics. Information concerning your family history will not be shared with others unless you provide us with permission to do so. In some instances, we request that patients to sign a "Release of Information" form prior to releasing medical records.

Q. What can I expect during my first visit to genetics?

- A. Appointments normally last between one and two hours. During that time you will meet with a Genetic Counsellor and/or Medical Geneticist who will review your/your child's medical and family history. If a physical exam is needed it will be done by the Medical Geneticist. Following that the Genetic Counsellor and/or Medical Geneticist will review with you the findings and the recommended testing, the possible diagnosis and the implications, and suggest plans for follow-up.

Q. Will I (my family) be given a specific diagnosis?

- A. All of your doctors are making every attempt to provide you with a diagnosis or to do testing to confirm a diagnosis that has been previously established. However, it is important to remember that we are not always able to provide a specific diagnosis or offer testing for a specific disease. Genetics is a field that is rapidly advancing but there is still much to discover.

Q. Do I have to be tested? Will the Genetic Counsellor tell me what to do?

- A. The decision to have genetic testing is personal. No one can tell you what you should or should not do. We will explore the benefits and limitations of any test with you so that you can decide whether or not testing is right for you and your family.

Q. What information will I receive at the appointment?

- A. We attempt to provide written resources to all of our patients. Many genetic conditions are very rare so this is not always possible. Please feel free to ask for a copy of the visit letter or for a family letter for you to share with other family members.

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