THE DIVISION OF MEDICAL GENETICS

Royal University Hospital, Saskatoon, Saskatchewan

FAQs FOR PHYSICIANS

Q. What is Medical Genetics?

A. Medical Genetics is the medical discipline concerned with the diagnosis, intervention and prevention of inherited conditions. Diagnosis of genetic conditions involves physical examination and laboratory blood work including chromosome analysis, DNA tests and/or biochemical analysis. If a diagnosis is made we discuss the inheritance and management of the condition and refer patients to other specialties as necessary. In working with our patients we also focus on the psychosocial impact of the problems they are dealing with and assist them in making the best decision for their family.

Q. When should I refer to Medical Genetics?

A. Often the most appropriate time to refer patients is 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 differences 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 can my patients expect to gain from a Medical Genetics referral?

A. Genetics professionals attempt to identify an underlying cause for the health and developmental concerns in a family. With the information, investigations, and diagnoses obtained by other healthcare professionals and ourselves, we teach the patient about the condition, the resources available to them and the appropriate follow-up. We provide strategies for sharing health information between family members.

Q. What is the referral process?

A. Referrals can be mailed or faxed to the Division of Medical Genetics. Once a referral is received it is triaged using the information that is provided to us. For example, prenatal patients are seen within days of a referral, however there is a waiting list for most other referrals unless an urgent medical need has been identified. In those cases, appointments are booked once family history forms have been completed and returned. Appointments are generally not booked prior to having the family history forms completed because these family history forms not only contain valuable information but they also are an indicator of the patient's intent to attend a scheduled appointment. If a physician believes that a referral needs to be prioritized this should be clearly stated on the referral letter or the case discussed with one of the members of the medical genetics team.

Q. What information is needed to ensure a referral is triaged appropriately?

A. We require the patient's demographic information including: Name, DOB, address, phone number(s), and the reason for referral. It is very helpful for us to know who else in the family has received a similar diagnosis or is experiencing similar symptoms. Family charts and pedigrees are maintained in the department. Please let us know the names of any family members previously referred to or seen by Medical Genetics. Additional information on whether other family members are living or deceased and the place of diagnosis or treatment are extremely useful in allowing us to confirm a diagnosis and provide accurate counselling. Results of any related testing (e.g., CK in muscle disorders) should also be included.

Q. Who will my patient see?

A. The patient will meet first with a Genetic Counsellor who will review the patient's medical and family history. If a physical exam is required the patient will be examined by a Medical Geneticist. Counselling is then provided by the Medical Geneticist and/or the Genetic Counsellor. We will review findings from the appointment, recommend testing, discuss the possible diagnoses and implications, and make plans for follow-up. Appointments normally last between one to two hours.

Q. What resources will be provided?

A. We attempt to provide families with written information on the condition including information on the inheritance pattern. We offer information on local or national support groups and organizations. In addition, we can try to match families with others experiencing similar situations. We also provide information to help patients identify family members for whom an evaluation or testing is indicated/appropriate.

Q. Why would I refer if a diagnosis will not change management?

A. Often a genetic evaluation can answer questions that the family may have. Questions such as: Why did this happen? Will it happen again? Why are there so many medical concerns in our family? Sometimes this is as important to a family as knowing what they can do about it.

Q. My patient had genetic testing but the results were normal. What should I do now?

A. Many genetic conditions are still diagnosed on a clinical basis only. This is because not all genetic disease has been linked to a specific gene. As well, many genetic tests have technical limitations and many are still in the research stage. Discussion among health professionals is needed to accurately diagnose and consistently follow these individuals. Encourage the patient to contact the Medical Genetics Clinic periodically to see if further testing is available.

Q. How does ethnic background impact genetic risk?

A. The chance of being a carrier of certain genetic disorders varies among people from different ethnic backgrounds. Specific carrier testing should be routinely offered to individuals of certain ethnicity. For example, there is an increased risk for Tay-Sachs disease in the Ashkenazi Jewish population. Carrier testing prior to pregnancy allows individuals to receive the appropriate counselling and ensures that timely prenatal diagnosis can be offered to at-risk couples.

Q. Why would I refer a patient for prenatal genetic counselling if she would not consider abortion?

A. It is every patient's right to gain as much information about her pregnancy as possible. Patients use this information in many different ways. Choices include using the information to be more informed and better prepared when a baby is born with health concerns. This offers parents time to adjust and to develop coping strategies if the baby should have a serious medical condition. Prenatal diagnosis allows parents a chance to meet with the physicians and surgeons who may be involved in the child's care following birth and to have time to make decisions about how much effort should be made to maintain a child's life. Parents should also be aware of the opportunity to put the child up for adoption if they do not feel that they will be able to cope with ongoing medical needs. If serious anomalies or a lethal condition is diagnosed prenatally, some families may appreciate the opportunity to re-consider terminating the pregnancy.

Q. What role does genetics play in the evaluation of infertility?

A. Approximately 15% of couples trying to achieve a pregnancy experience infertility. There are a number of causes of infertility including structural anomalies of the reproductive tract in men and women, hormonal imbalances, chronic infections, environmental factors, chronic disease and genetic factors such as cytogenetic anomalies, Y chromosome microdeletions, and genetic disease such as cystic fibrosis (CF) causing congenital absence of the vas deferens, either bilateral or unilateral (CBAVD or CUAVD). A genetics evaluation may help elucidate a cytogenetic or molecular cause of infertility, allow discussion of the psychosocial issues surrounding infertility and the risks associated with assisted reproduction.

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