

# **A Guide for Parents and Prospective Parents Seeking Genetic Counselling**



**Canadian Institute of Child Health**

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## About This Guide

The purpose of this guide is to increase your understanding and readiness to take advantage of genetic counselling services.

You may find the guide useful if you are a prospective parent who:

- during the course of prenatal testing, has received abnormal results
- was born with a physical difference such as, for example, cleft palate, heart malformation or club foot
- has a close relative with a physical difference or health condition that may be genetic
- has had recurrent unexplained pregnancy losses (three or more miscarriages) or lost children at birth or in infancy
- is older than 40 years of age
- is concerned about genetic conditions that are frequent in some ethnic groups (e.g., Thalassaemia in people of Mediterranean and Southeast Asian descent, Sickle-Cell Disease among individuals of African or Caribbean ancestry, or Tay-Sachs Disease in Jewish persons of Eastern European descent)
- is concerned about the possible effects of being exposed to radiation, drugs and alcohol, chemicals, or infections

This guide was written by Sylvia Reitmanova, based on information contained in the Genetics and Paediatric Health Module developed by Canadian experts in the field of genetics and paediatric health under the umbrella of Canadian Institute of Child Health.<sup>1</sup> We thank the Canadian Association of Genetic Counsellors, Drs. David Dymant and Gail Graham for their review of early drafts and endorsement of this guide.

<sup>1</sup> Canadian Institute of Child Health. <https://cichprofile.ca/module/2/>

## A, B, Cs of Genetics and Genetic Conditions

All biological parents transfer to their offspring genetic material that will influence their biological characteristics. This information is contained in small heritable units, called genes. Genes are composed of a chemical called DNA (deoxyribonucleic acid) [pictured below]. Each person has about 20,000 genes grouped together into physical structures, called chromosomes. Human cells have 23 pairs of chromosomes; one set from each parent.



Changes in a single gene or chromosome numbers can be responsible for the development of a genetic condition. Examples of genetic conditions include cystic fibrosis, Duchenne muscular dystrophy, or Down syndrome. Gene mutations can be passed from parents to their offspring or they may happen out of the blue, usually in a single egg or sperm that conceives a baby. Some mutations are thought to result from exposure to harmful factors such as radiation, chemicals, or infections prior to, or during, pregnancy.

Some genetic conditions, such as Down syndrome or spina bifida, can be detected by screening during pregnancy. Prenatal screening typically involves maternal blood tests and ultrasound examination. Most provincial and territorial health insurance programs cover prenatal screening. Prenatal screens are not diagnostic tests – they simply indicate that a child is at risk for certain conditions. Diagnostic genetic testing by chorionic villus sampling or amniocentesis is typically necessary to confirm a prenatal screening result.



Participation in prenatal screening is an individual choice of prospective parents based on their values and experiences. Some will want to know if their pregnancy is at risk for a genetic condition, or affected by a genetic condition, in order to prepare to care for a child with a genetic condition. Others may want the information to help them decide whether to carry on with a pregnancy or not, while others may want information to plan for future pregnancies. The results of screening may cause mixed emotions, including stress, anxiety, relief, guilt, and questions about with whom to share the information. Professional genetic counselling can provide comprehensive support to parents during this challenging time.

Some prospective parents may feel tempted to buy genetic tests provided by private commercial companies operating on-line. Unless these tests are ordered directly by healthcare professionals, genetic counsellors do not recommend parents use them because they can be unreliable and are offered without proper clinical indication. Also, it is not guaranteed that individuals undergoing private commercial testing will have access to a healthcare professional who can offer them appropriate genetic counselling after they receive testing results.



## Genetics Counselling Services

If you have concerns about the risk of passing a genetic condition to your child because you have a family history of concern or your prenatal screening indicates a potential genetic condition, or you were exposed to harmful factors that could affect your developing fetus, you may find it useful to request a referral to a genetic counsellor. You may also locate and contact a genetic clinic directly. Their list is available on the website of Canadian Association of Genetic Counsellors.<sup>2</sup>



<sup>2</sup> Canadian Association of Genetic Counsellors. <https://www.cagc-accg.ca/>

A genetic counsellor is a specially trained health professional who works in a team with medical geneticists and other healthcare professionals who can help you to:

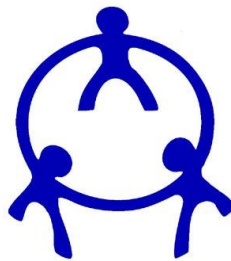
- examine your medical and family history to evaluate your risk of developing, or having a child with a hereditary condition
- understand how genetic conditions may be passed down through families
- interpret the results of genetic testing, explain how a genetic condition could affect the health and well-being of your child and describe the challenges that children and parents may face, including the issues of stigmatization or discrimination
- understand your options and what treatment and supports are available
- make informed decisions, taking into account your risk, your family goals, and your ethical and religious beliefs in addition to exploring ways to communicate the information to other family members
- access other support and advocacy groups for additional resources

Working with genetic counsellors can be a rewarding experience. They will provide you with the information and support which can reduce your stress or anxiety to navigate the challenges associated with the risk of genetic conditions. Talk to your primary care provider if you think you would benefit from genetic counselling.

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<sup>1</sup> Canadian Institute of Child Health. <https://cichprofile.ca/module/2/>



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