Genetics and Paediatric Health

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Contributors:

Writing and editing:
Denise Avard
Jennifer Fitzpatrick
Louise Hanvey
Cheryl Rockman-Greenberg

Project development, research, and review:
Expert Advisory Committee
Laura Arbour
Denise Avard
Pascal Borry
Pranesh Chakraborty
Jennifer Fitzpatrick
William Fraser
Jan Friedman
Louise Hanvey
Anne Junker
Bartha Maria Knoppers
Elaine Orrbine
Cheryl Rockman-Greenberg
Jacques P. Tremblay
Brenda Wilson
Durhane Wong-Rieger

Other
Denise Alcock
Jeannine Fraser
Aspasia Karalis
Meghan Marcotte
Kelly McClellan
Vaso Rahimzadeh
Karine Sénécal
Janice Sonnen
Robin Walker

Graphic design:
Shelley Callaghan
Louise Hanvey
Meghan Marcotte
Bert Schopf

Website development:
Accel Web Marketing

Canadian Institute of Child Health
Institut canadien de la santé infantile
McGill
University of Manitoba
Winnipeg Regional Health Authority
Caring for Health
Office régional de la santé de Winnipeg
À l'écoute de notre santé

1.1.1 About Genetics and Children's Health: Some Biology

Image credit: Shutterstock

An individual's genetic make-up forms what is known as the genome. The human genome is made up of nearly 20,000 genes. Though specific genes control specific parts of human development and health, they are grouped together into physical structures called chromosomes and are located in the nucleus of each cell. Humans have 23 pairs of chromosomes, so every individual has two copies of every gene and a child receives one copy of each gene from each parent. A gene is made of deoxyribonucleic acid (DNA), which is the genetic instruction manual.

There are a number of websites where you can find more information about the genome, for example:

- SickKids, the Genetics section (If a user profile box pops up, you can press 'cancel' or create a user profile to access this site.)
- Utah Genetics Education, Tour of Basic Genetics

1.1.2 Genes and How They Work

Image credit: Shutterstock

Genes provide the directions for building every molecule the human body requires. There are hundreds of different kinds of cells in the body, and it is the variety in gene function and expression that determines these differences.

Gene expression is the process whereby the information in a gene produces the final gene product – a protein. Gene expression refers to genes being turned "on'" or "off" – that is, stimulated to produce protein. Healthy development of all organs in the body depends on which and when certain genes are expressed. You can think of genes acting as an orchestra, turning on and off, and producing proteins that act together – in concert-like fashion – in the development of an individual. Many factors can influence the individual instruments (genes) in this complex orchestra.

Research shows that environmental factors and early experiences have the power to chemically influence gene expression and hence control their function.

There are a number of websites where you can find more information about genes, for example:

- Utah Genetics Education, the Epigenetics section
- DNA Learning Centre

2.1.1 Chromosonal Conditions

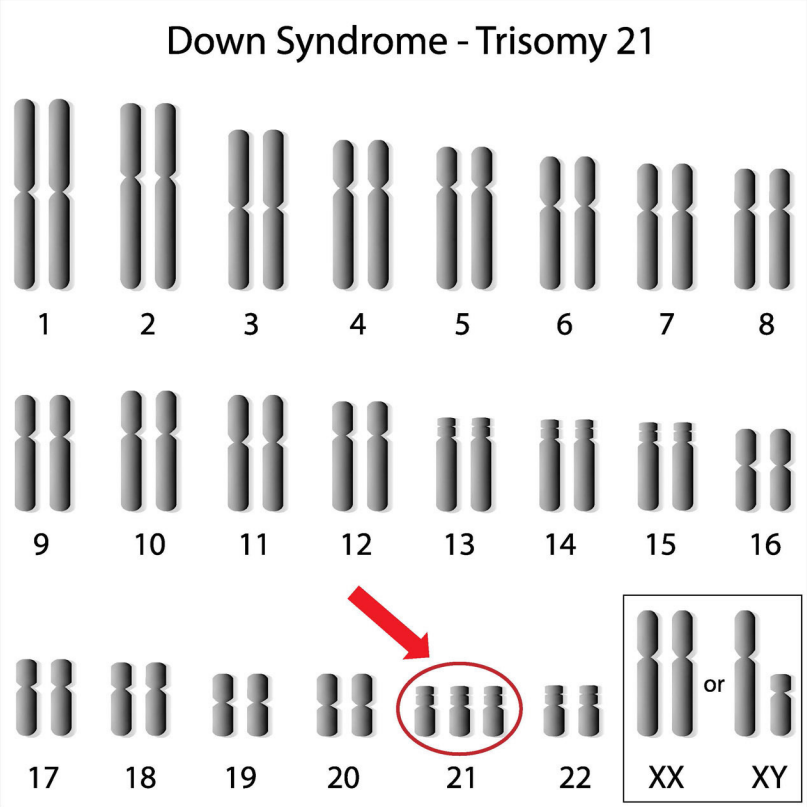


Image credit: Shutterstock

Chromosomal conditions or anomalies occur when there is an abnormal amount of chromosomal material. The usual chromosome complement in each cell is 46. There are 22 pairs of autosomes numbered 1 to 22, with each pair having the same genes. The 23rd pair is called the sex chromosomes, with females having two "X" chromosomes and a male having an "X" and a "Y" chromosome. The "X" and "Y" chromosomes, unlike autosomes, have different genes. There is sometimes an abnormal total number of chromosomes – sometimes an extra chromosome. For example, a child with Down syndrome has three copies of chromosome 21, rather than the usual two, so the condition is called "trisomy 21."

You can find more information about these conditions and how they happen at:

- SickKids, the Genetics section (If a user profile box pops up, you can press 'cancel' or create a user profile to access this site.)
- Centers for Disease Control and Prevention, Facts on Pediatric Genetics

2.1.2 Single Gene (Monogenic) Conditions

Image credit: Shutterstock

Single gene conditions (also called monogenic conditions) are caused by abnormalities, or mutations, of a single gene. These conditions can be severe or harmless, and though most are rare, some can be common. Familial hypercholesterolemia, a genetic condition that can lead to high levels of a harmful form of cholesterol, is an example of a common single gene condition.

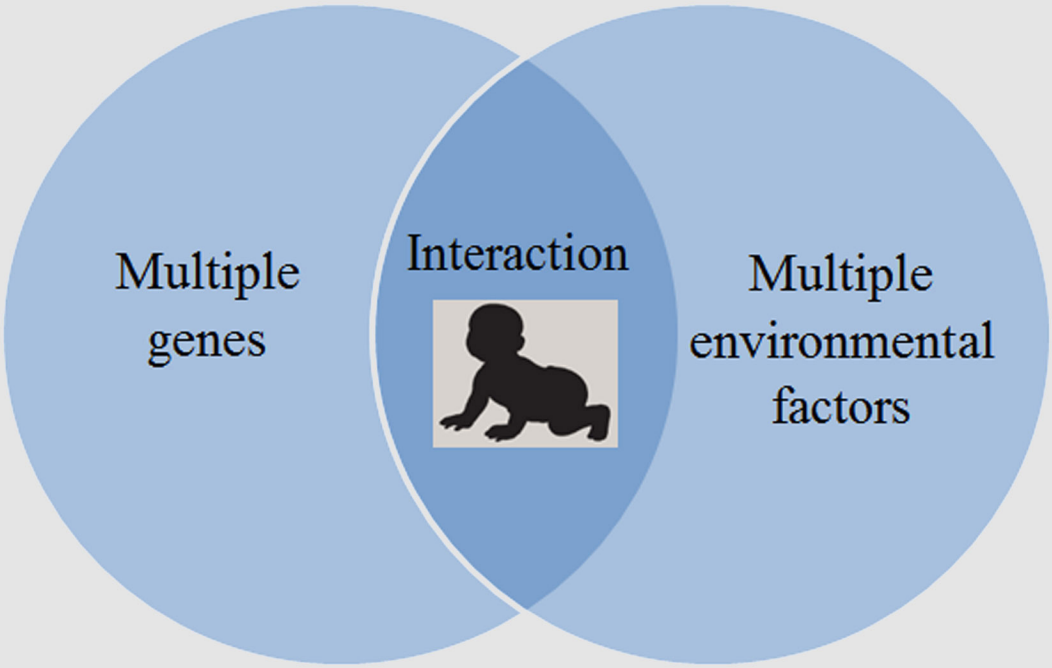
Single gene conditions can have either a dominant or a recessive inheritance pattern. Familial polyposis is a single gene condition that results from a dominant mutation. Dominant inheritance means that the condition occurs when only one copy of a gene pair has a mutation. Familial polyposis often results in cancer of the large intestine (colon) and rectum.

Other single gene conditions only develop when the person inherits two mutations in the same gene on one of the autosomes – one from each parent. These mutations are known as autosomal recessive. An individual with only one copy of a recessive gene mutation does not develop the disease and is instead called a carrier. Some examples of autosomal recessive, single gene conditions are cystic fibrosis, Tay-Sachs disease, and sickle cell anaemia. In these conditions two copies of a mutation in the gene – one from each parent – must be present for a child to develop the condition.

When a mutation exists in a gene on the X chromosome, an X-linked condition may result. An example of an X-linked condition is hemophilia.

For more information about these conditions and how they happen go to: Utah Genetics Education, National Coalition of Professional Education in Genetics, and Genetic Alliance UK.

2.1.3 Multifactorial Disorders or Complex Traits



Graphic created by CICH using a Shutterstock image.

Multifactorial genetic disorders, or complex traits, result from several genes in combination with lifestyle and environmental influences. Multifactorial disorders include diseases like diabetes, many cancers, heart disease, and asthma. Multifactorial disorders account for the largest group of genetic conditions, both in numbers and the impact that they have on child health and the health care system as a whole.

For more information about these conditions and how they happen go to:

- Centers for Disease Control and Prevention, Facts on Pediatric Genetics
- National Coalition of Professional Education in Genetics
- University of Kansas, Genetics Education Center
- Centre for Genetics Education

2.1.4 What Causes Genetic Conditions?

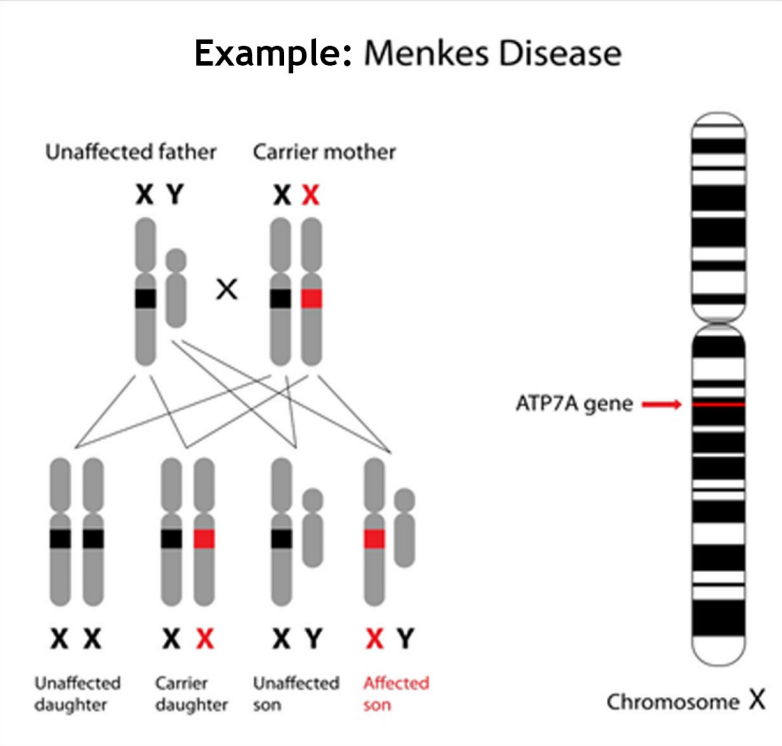


Image credit: Shutterstock

Menkes disease is a recessive genetic condition caused by a mutation of the gene on the X chromosome that is responsible for the metabolism of copper in the body. Copper levels become abnormally low in the liver and brain and abnormally high in the kidney and intestinal lining. Menkes disease leads to premature birth, floppy muscle tone, developmental delay, seizures, and failure to gain weight and to grow at the usual rate.[1,2,3]

Gene mutations can be inherited from parents; however, mutations in genes and chromosomes can happen unexpectedly and usually for unexplained reasons. Mutations that exist in the parents' genes are inherited mutations. New mutations, which occur during the transmission of genetic material to the next generation, are called de novo mutations. It is important to obtain a complete family history to better understand genetic risk factors for children and their families.

For more information about inheritance see:

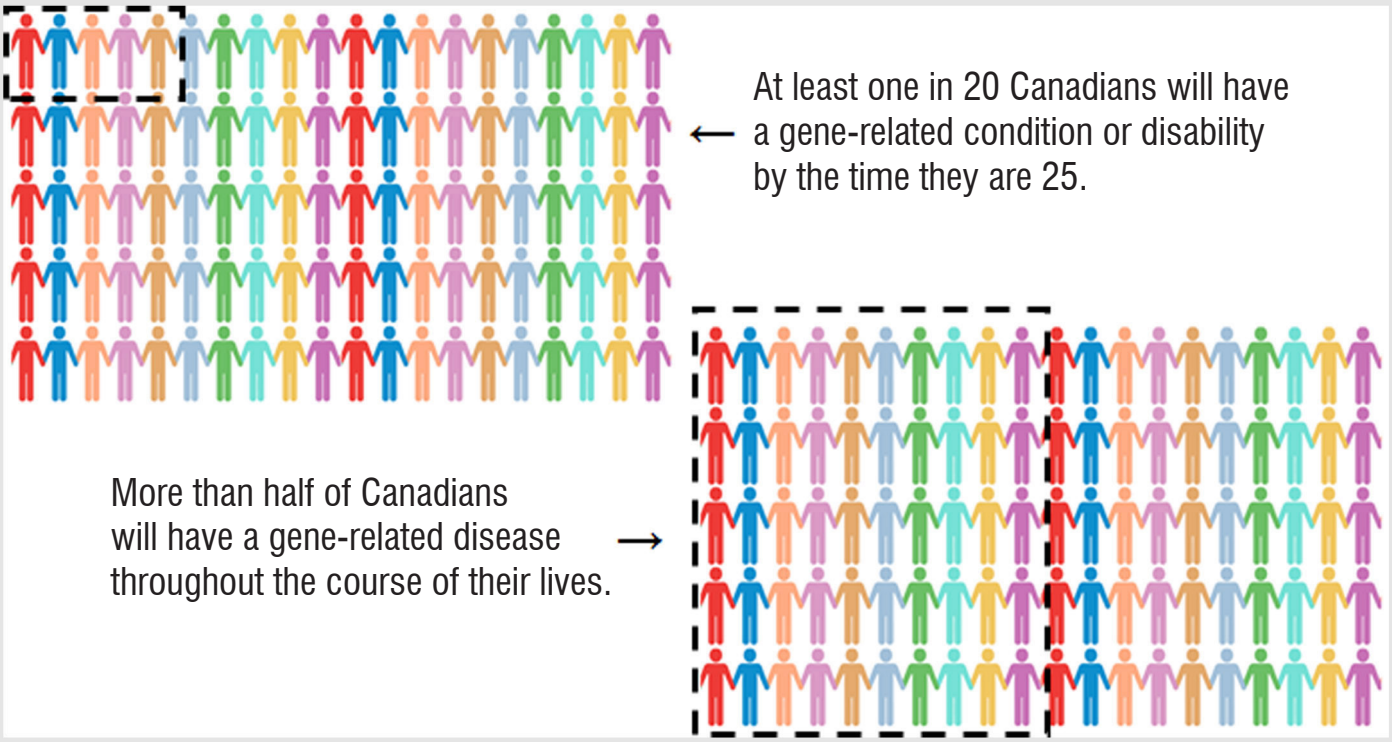
- SickKids, 'Genetics' section (If a user profile box pops up, you can press 'cancel' or create a user profile to access this site)
- Centers for Disease Control and Prevention, 'Pediatric Genetics' section

1 http://ghr.nlm.nih.gov/condition/menkes-syndrome;

2 http://www.merriam-webster.com/medical/menkes'%20disease;

3 http://www.ninds.nih.gov/disorders/menkes/menkes.htm

2.1.5 How Many Children Have Genetic Conditions?



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In Canada, it is estimated that at least one in 20 individuals will experience a gene-related condition or disability by the age of 25. In addition, more than half the population will experience a gene-related disease during their lifetime.[1]

[1] Science Council of Canada. Genetics in Canadian Health Care. Report 42. Ottawa: Science Council of Canada; 1991

2.1.6 What Does this Mean for Children and Families?

Image credit: Shutterstock

When a genetic diagnosis is suspected or confirmed, the child and his or her family members often require complex support and care, perhaps for a significant period of time. Parents will have questions about what the condition will mean for their child and also how the condition originated, either through genetic inheritance or through a spontaneous genetic mutation, what the risks are of it happening again or in future generations, and what their reproductive choices might be for another pregnancy. They may be concerned about their health and the health of their child, privacy, stigma, or discrimination, and parents often experience guilt.[1, 2] Often other relatives are concerned about themselves or their children. Because there is no cure or effective treatment for many genetic conditions, the disclosure of a genetic diagnosis can be incredibly difficult for all family members involved.

Personal, family, cultural, and religious beliefs influence how families interpret and share information about genetic conditions. There has been an explosion of knowledge and information regarding the genetic bases of disease. It is therefore critical to ensure appropriate clinical services are available to families who need them.

[1] Wilcken B. Ethical issues in genetics. Journal of Paediatrics and Child Health.2011;47:668–71.

[2] Canadian Association of Genetic Counsellors, https://cagc-accg.ca/content/view/12/26/

3.1.1 What Genetics Services Do Children and Families Need?

Image credit: Shutterstock

Since genetics is a rapidly evolving clinical field, patients and their families are confronting increasingly complicated issues in their healthcare. As such, a family recently diagnosed with a genetic disorder requires both medical and emotional support.

Genetic diagnoses are made by physicians trained to recognize signs and symptoms of genetic diseases, and skilled in interpreting genetic test results. They are medical geneticists.

Many other physicians – both generalists and specialists – are also able to establish genetic diagnoses as a result of a family's history, signs, and symptoms.

Genetic counsellors are specially trained health professionals who help address issues before and after a person decides to take a genetic test. They are an important resource for families seeking to understand genetic information.

3.1.2 What Genetic Services Exist for Children and Families in Canada?

87*
Medical Geneticists[1]

269
Genetic Counsellors[2]

* This number reflects the medical geneticists working at 16 of the 17 academic paediatric health centres in Canada. Medical geneticists working at the Northern Ontario School of Medicine or in non-academic paediatric health centres were not included in this count.

Graphic created by CICH using Shutterstock images and data from:

[1] Pediatric Chairs of Canada (PCC), Academic Workforce Survey, 2012; and

[2] Canadian Association of Genetic Counsellors, Membership directory. Oakville, Canada, 2013.

Genetic services are organized through genetic clinics across the country that employ interprofessional teams of healthcare providers. Two important providers on this team are medical geneticists and genetic counsellors. There are approximately 87 medical geneticists in Canada; most practice in British Columbia, Alberta, Ontario, and Quebec, but they are found in other provinces as well. The majority are in academic centres, but they provide outreach to smaller cities, as well as rural and remote areas.

In Canada, the Canadian Association of Genetic Counsellors certifies genetic counsellors. There were approximately 269 certified genetic counsellors in Canada in 2009.

Alongside these genetic services, there are a number of community-based organizations in Canada that focus on providing support and education to families with a genetic disorder. The Canadian Directory of Genetic Support Groups lists 78 Canadian organizations offering such support. The umbrella organization, Canadian Organization for Rare Disorders (C.O.R.D.) provides overarching support and advocacy for many genetic disorders.

Of the genetics services available, laboratory services are a central component. All academic pediatric centres offer cytogenetic, molecular, and/or biochemical laboratory services; however, these vary from centre to centre.

3.1.3 Who Might Be Seen by a Medical Geneticist?

Image credit: Shutterstock

Medical geneticists are physicians who, after finishing their medical degree, receive specialist training in genetics through one or more of the following: the Canadian College of Medical Geneticists, Collège des médecins du Québec, or Royal College of Physicians and Surgeons of Canada.

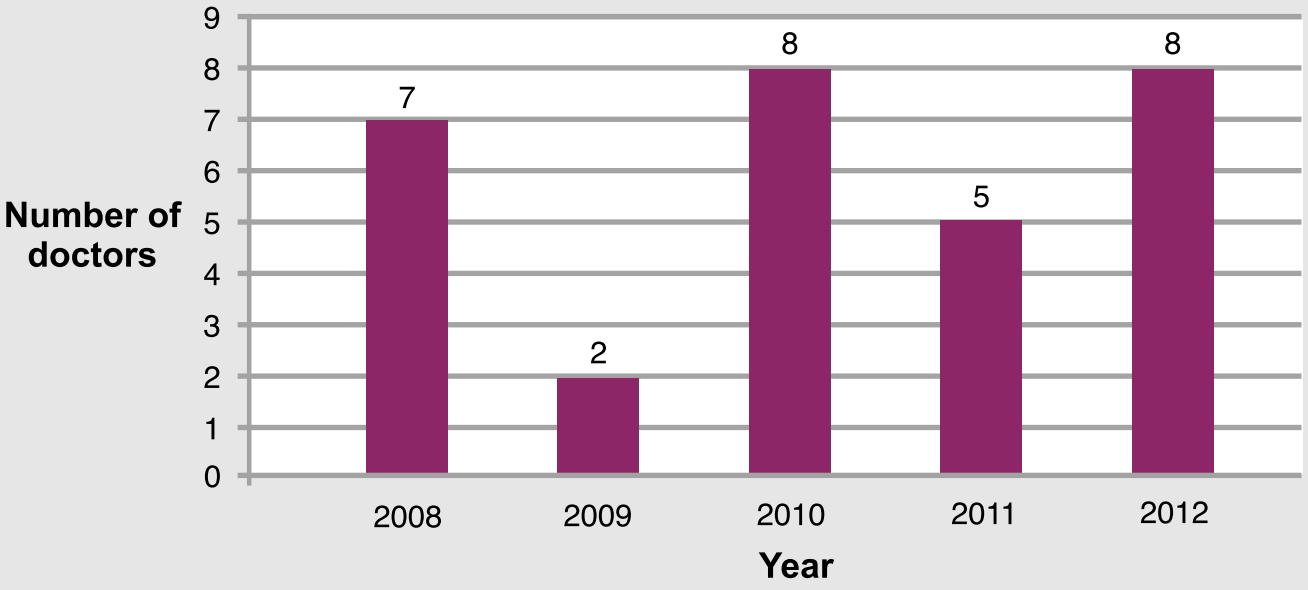
Medical geneticists assess, diagnose, and counsel individuals and their families who may have a genetic condition. Many medical geneticists provide direct care to children and adults with genetic conditions. Although most genetic conditions do not have a definitive treatment, such options are expanding greatly over time.

Who might be seen by a medical geneticist?

- Individuals with a known or suspected genetic condition.
- Family members of those with a known or suspected genetic condition.
- Children with different facial or body features, congenital anomalies, developmental challenges, learning difficulties, or a combination of these conditions.
- Individuals with a family history of cancer, such as breast or bowel cancer.
- Couples with a history of recurrent miscarriages.
- Couples or families following the death of a child from a known or suspected genetic condition.
- Couples for whom an abnormality with potential genetic implications has been detected during one or more pregnancies.

BMJ Careers; http://careers.bmj.com/careers/advice/view-article.html?id=158

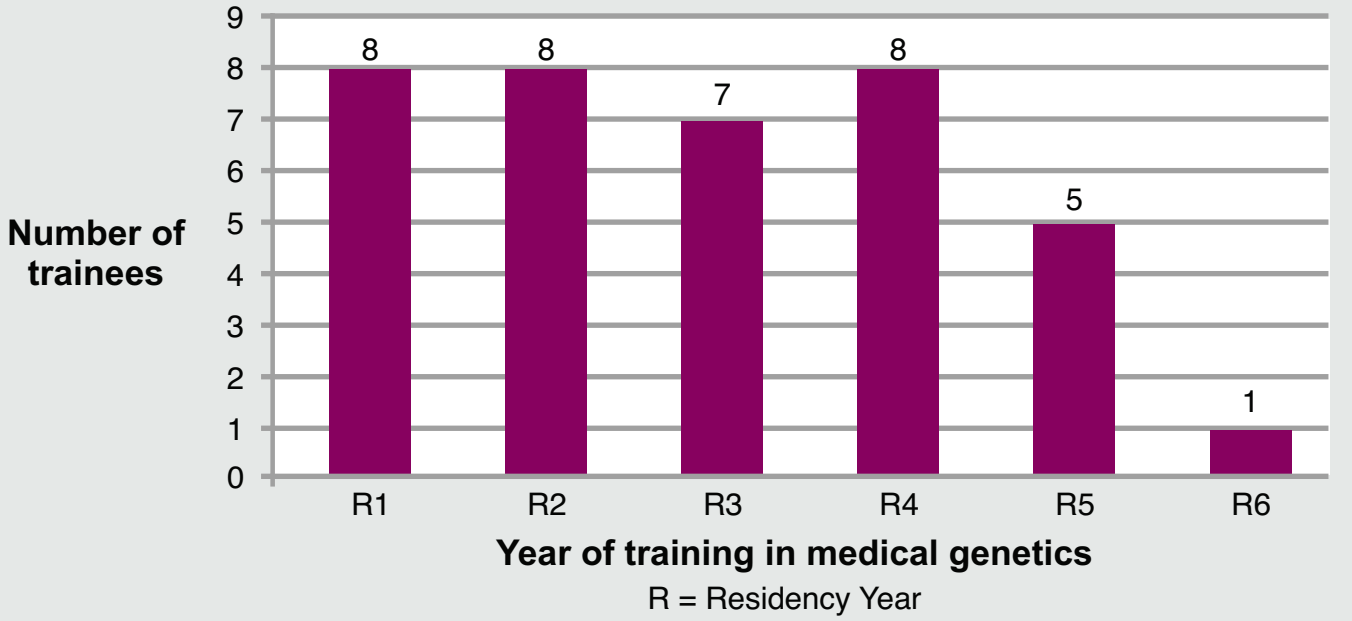
3.1.4 The Number of Doctors Who Completed Specialty Training in Medical Genetics, Canada, 2008 to 2012



Graphic created by CICH using data from CAPER. Annual Census of Post-MD Trainees. 2012-2013. http://caper.ca/~assets/documents/pdf_2012-13_CAPER_Census.pdf

A small number of doctors receive specialty training in medical genetics each year in Canada. Between 2008 and 2012 (inclusive) 30 doctors completed specialty training in medical genetics. The number of doctors who completed this training per year varied from 2 to 8.

3.1.5 The Number of Post-Graduate Trainees in Medical Genetics, by Year of Training, Canada, Academic Year 2012-2013



Graphic created by CICH using data from CAPER. Annual Census of Post-MD Trainees. 2012-2013. http://caper.ca/~assets/documents/pdf_2012-13_CAPER_Census.pdf

In 2012/13, there were 37 post-graduate trainees in medical genetics in Canada. They are relatively equally distributed across the first five years of training.

CAPER. Annual Census of Post-MD Trainees. 2012-2013. http://caper.ca/~assets/documents/pdf_2012-13_CAPER_Census.pdf

3.1.6 Genetic Counsellors

Image credit: Shutterstock

Genetic counsellors have specialized training and experience in medical genetics and counselling. In Canada, genetic counsellors have a Master of Science degree in genetic counselling from a recognized university program. In Canada, the Canadian Association of Genetic Counsellors certifies most genetic counsellors.[1]

Genetic counsellors provide counselling support so that people can make informed decisions about their health and the health of their children. They usually work in genetic clinics. They also serve as educators and resource people for other healthcare professionals and for the general public.[2]

[1] Canadian Association of Genetic Counsellors, http://www.cagc-accg.ca/

[2] Canadian Association of Genetic Counsellors. What is a Genetic Counsellor? Oakville, Canada: Canadian Association of Genetic Counsellors. http://www.cagc-accg.ca/doc/what%20is%20a%20genetic%20counsellor%20-%20english.pdf

For more information about genetic counsellors and genetic counselling see:

- What is a Genetic Counsellor? by the Canadian Association of Genetic Counsellors.

3.1.7 Genetic Counselling

Image credit: Shutterstock

Genetic counselling involves communicating with patients and families about the issues that arise when someone has a genetic condition, or if there is risk of a genetic condition in a family. Genetic counsellors help an individual or family to:

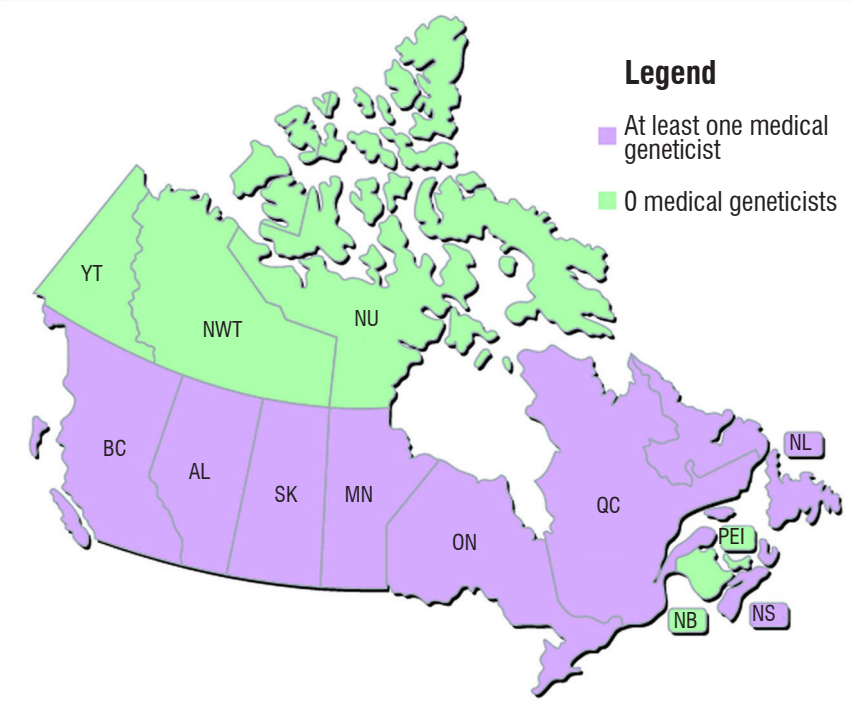
- Understand the medical facts, including a child's diagnosis, how the condition will affect his/her health and wellbeing, and what treatment and support are available.
- Review their family and medical histories.
- Understand how genetic conditions are passed down through families.
- Determine if genetic testing is available for a genetic condition.
- Understand the risks associated with the condition.
- Understand the options for dealing with a genetic condition so they can make informed decisions regarding genetic testing.
- Understand the latest research or treatment information.
- Decide on the best course of action for the family in view of their risk, family goals, and ethical and religious standards.
- Discuss the best possible treatment options for the condition that also considers the risk of recurrence for the disorder.
- Obtain referrals to medical specialists, advocacy and support networks, and other resources and support groups.[1,2,3]

[1] Canadian Association of Genetic Counsellors. What is a Genetic Counsellor? Oakville, Canada: Canadian Association of Genetic Counsellors. https://cagc-accg.ca/images/docs/GC_Career_Brochures/what%20is%20a%20genetic%20counsellor%20-%20english.pdf

[2] Fraser FC. Genetic Counseling. American Journal of Human Genetics. 1974;26:636–61; cited in Leeming W. Looking Back on the Future of Genetic Counselling in Canada. CBMH/BCHM. 2013;30:1.

[3] Johns Hopkins National Human Genome Research Institute. Making Sense of your Genes: A Guide to Genetic Counseling. Chicago: National Society of Genetic Counselors, Inc. and Washington, DC: Genetic Alliance; 2008. http://www.kumc.edu/gec/prof/guidetogc.pdf

3.1.8 Access to Services - Medical Geneticists



Province/ Territory	Number of geneticists at university health centres(1)	Geneticists at university health centres(1) /100,000 population (0–18 year olds)
AB	14	0.9
BC	6	0.71
MB	5	1.81
NB	-	-
NL	2	2.02
NT	-	-
NS	6	1.66
NU	-	-
ON	32	8.86
PE	-	-
QC	21	1.28
SK	1	0.43
YT	-	-
TOTAL	87	1.19

Sources: Statistics Canada. CANSIM table 051-0005. Estimates of population, Canada, provinces and territories, quarterly, July 1, 2013. Ottawa: Statistics Canada; 2013. http://www5.statcan.gc.ca/cansim/a26?lang=eng&retrLang=eng&id=0510005&paSer=&pattern=&stByVal=1&p1=1&p2=31&tabMode=dataTable&csid= and Pediatric Chairs of Canada (PCC), Academic Workforce Survey; 2012

Within 16 academic paediatric health centres across Canada there are 87 medical geneticists.[1] Access to these medical geneticists and the services they provide is not equitable across the country. For example, in Saskatchewan there is less than one medical geneticist per 200,000 children and youth up to 18 years old. Within Ontario, this number has reached close to nine medical geneticists per 100,000 residents. Thus, in Ontario the ratio of medical geneticists to 0–18 year olds is more than 20 times greater than that in Saskatchewan.

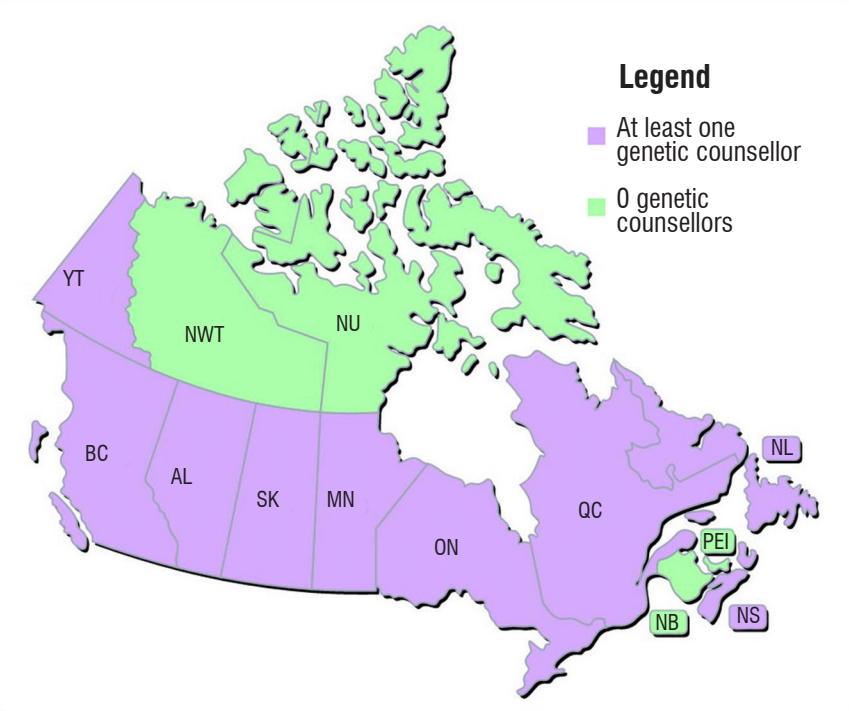
[1] This number reflects the medical geneticists working at 16 of the 17 academic paediatric health centres in Canada. Medical geneticists working at the Northern Ontario School of Medicine or in non-academic paediatric health centres were not included in this count.

Implications

Lack of access to genetic clinics and medical geneticists can result in waiting lists and wait times. Wait times vary tremendously across the country. In some instances waiting times can even be greater than a year.[2] Long waits such as these may delay the identification of genetic or other medical conditions as well as timely intervention. In many cases, this delay may lead to more negative outcomes of the condition and greater costs associated with its treatment. Additionally, the time and cost required to travel to health centres either far away within one's own province or territory or to a different province or territory adds additional challenges for many families, children, and youth to get the care they need. As time passes, families and children are left with many unanswered questions and concerns.

[2] Nova Scotia Wait Time Information; Genetic Consultation. http://waittimes.novascotia.ca/procedure/genetic-consultation.

3.1.9 Access to Services - Genetic Counsellors



Province/ Territory	Genetic Counsellors	Population/ Genetic Counsellors (thousands)
AB	31	129.84
BC	46	99.61
MB	5	253
NB	-	-
NL	10	52.67
NT	-	-
NS	13	72.37
NU	-	-
ON	109	124.2
PE	-	-
QC	48	169.9
SK	6	184.72
YT	1	36.7
TOTAL	269	130.7

Sources: Statistics Canada. CANSIM table 051-0005. Estimates of population, Canada, provinces and territories, quarterly, July 1, 2013. Ottawa: Statistics Canada; 2013. http://www5.statcan.gc.ca/cansim/a26?lang=eng&retrLang=eng&id=0510005&paSer=&pattern=&stByVal=1&p1=1&p2=31&tabMode=dataTable&csid=; and Canadian Association of Genetic Counsellors. Membership directory. Oakville, Canada: Canadian Association of Genetic Counsellors; 2013.

There are 269 genetic counsellors in Canada.[1] This number includes all counsellors who are members of the Canadian Association of Genetic Counsellors – some of them serve specific populations. Access to these genetic counsellors and the services they provide is not equitable across the country. In British Columbia there are about 100,000 people per genetic counsellor. In Ontario that number is about 124,000; in Quebec, it is about 169,000; in Saskatchewan it is 184 000 and in Manitoba it is 253 000.

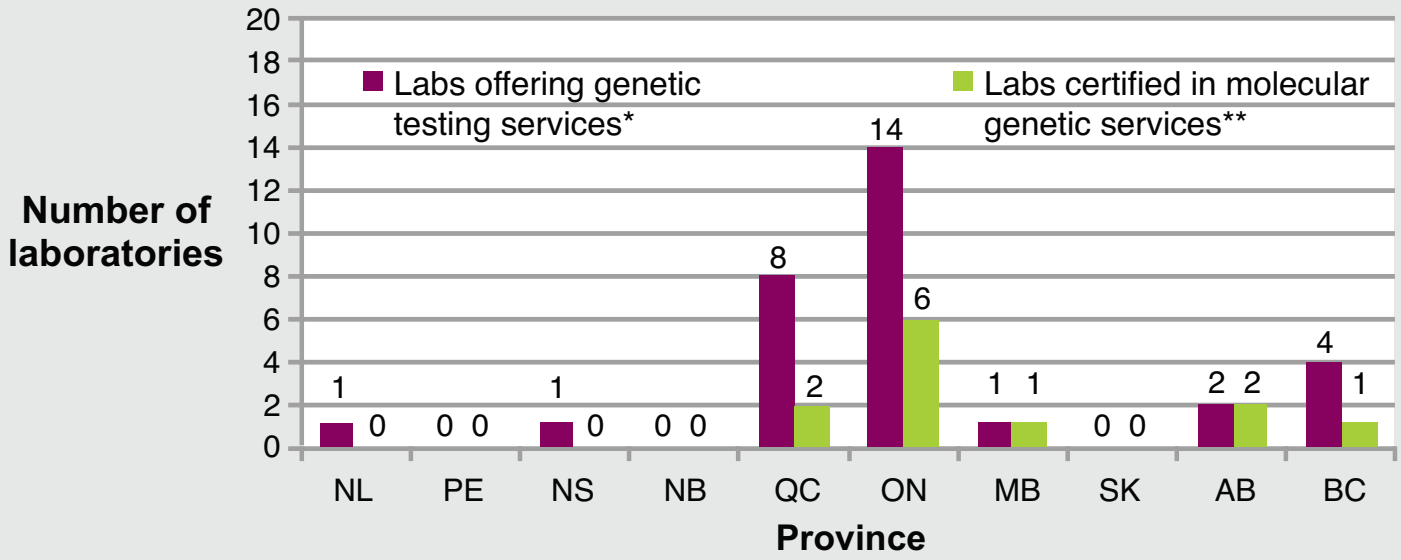
[1] Canadian Association of Genetic Counsellors. Membership directory. Oakville, Canada: Canadian Association of Genetic Counsellors; 2013.

Implications

Genetic counselling is an essential, integral part of healthcare for children and families with genetic conditions or those who may be at risk for genetic conditions. When a regional genetic clinic does not employ an adequate number of medical geneticists and genetic counsellors for the population, waiting lists can be very long. For many children, for example those with developmental delay, an early diagnosis and appropriate referral to treatment and support are essential to their optimal development.

Most genetic centres that employ genetic counsellors are in major cities. Therefore rural and remote regions of Canada may have limited access to genetic services except through outreach clinics and telehealth. We do not have data describing wait times for children and families, nor how many centres provide regular outreach, teleconferences or telehealth. It is critical that we develop data regarding services, wait times along with benchmarks that describe standards of the service and human resource mix.

3.1.10 Laboratories Providing Genetic Testing and Those That Are Certified Genetic Service Centres, Canada



* Number of laboratories offering genetic testing services (GeneTests web site www.genetests.org)
** Laboratories certified as a molecular genetic service centre by the CCMG (www.ccmg.medical.org)
† The data about labs offering genetic testing services is from 2014. The data about labs certified in molecular genetic services is from 2008.

Graphic created by CICH with data from: Somerville MJ, Allingham-Hawkins DJ. Regulation of Genetic Testing/Service in Canada, In: Kristoffersson U, Schmidtke J, Cassiman J, editors. Quality issues in clinical genetic services. Dordrecht, Heidelberg, London, New York: Springer; 2012. p.87.

Laboratories offering specialized genetic testing are accredited provincially. Each province has its own system. There are 35 laboratories that reportedly conduct genetic testing in Canada. Of these, 12 have certification from the Canadian College of Medical Geneticists as genetics service centres and are located in seven provinces. Since laboratory reporting is based on voluntary registration, these figures are likely to underestimate the number of certified laboratories.[1]

[1] For a list of the laboratories offering genetic services in Canada, view the Gene Tests website.

Implications

The administration of healthcare in Canada, which is provincially/territorially based, can create barriers to provinces/territories working cooperatively in the delivery of genetic testing.[1] Many centres do not have the funding to adopt the latest technologies to conduct tests as quickly as patients and their families might want. Further, the range of laboratory services available is not uniform across Canada.

Many factors influence decisions about whether or not genetic investigations are offered. These include the prevalence of the condition in the population and the validity of the test. They also need to include ethical, legal, social, and cost factors. It is often not easy to have rare diseases investigated.

[1] Somerville MJ, Allingham-Hawkins DJ. Regulation of Genetic Testing/Service in Canada. In: Kristoffersson U, Schmidtke J, Cassiman J, editors. Quality issues in clinical genetic services. Dordrecht, Heidelberg, London, New York: Springer; 2012. p.83–90.

3.1.11 Communication within Families

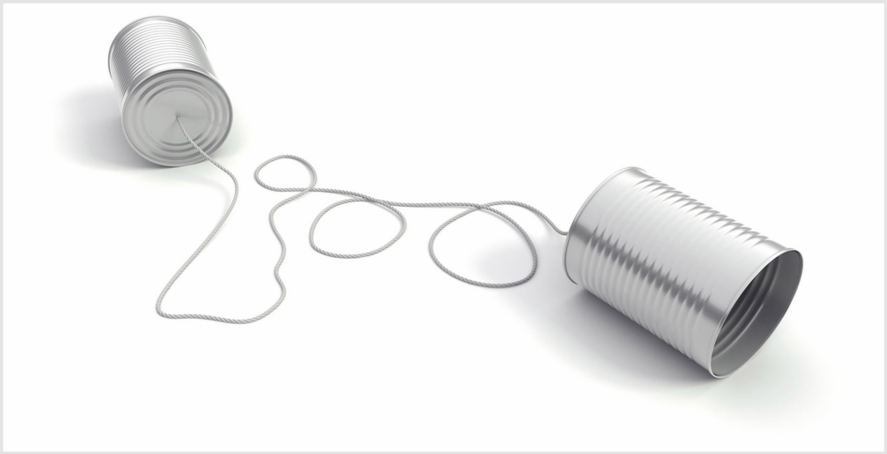


Image credit: Shutterstock

The result of a genetic test is often not only useful to the child and his or her immediate family, but also to the extended family. However, not all people are willing to communicate genetic information and test results to other family members. Communication within families is complex and delicate. It requires that the children and families who are affected must be able to absorb and understand complicated information about their own health. Oftentimes communicating a genetic diagnosis to other relatives relies on the degree of connectedness the individual maintains with relatives, among other things. Some patients may feel their diagnosis is a very private matter, while others may lack the connection needed with family members to communicate genetic results. Since physicians are bound by a duty to protect the privacy of their patients, the communication of sensitive genetic information to family members is placed directly on the patient's shoulders.[1,2]

Health professionals have a considerable role to play in guiding this type of communication within families. Age-appropriate information and personalized counselling are the cornerstones of effective genetic counselling for childhood genetic diseases.

[1] Nycum G, Avard D, Knoppers B. Intra-familial obligations to communicate genetic risk information: what foundations? what forms? McGill Journal of Law & Health. 2009;3:21–48.

[2] Godard B, Hurlimann T, Letendre M, Égalité N. Guidelines for disclosing genetic information to family members: from development to use. Familial Cancer. 2006;5(103):116.

Implications

Patients may be in the best position to anticipate the wishes of family members, who may or may not want to know about the genetic information in their family. For those patients willing to disclose, the healthcare professional's role in encouraging and supporting their patient's efforts to communicate with their family members is complex. In extremely rare life-saving situations, it is permitted for a physician to communicate genetic information to extended family members after all attempts to convince the patient to share such information have failed.[3] Better guidelines and policy for healthcare professionals with respect to counselling patients about communication within their families are much needed. These guidelines should include direction on how and when to involve children in these discussions, and may depend on many factors: age, comprehension, treatment availability or prevention for the disease, and recommended ages to initiate clinical screening or genetic testing.[4]

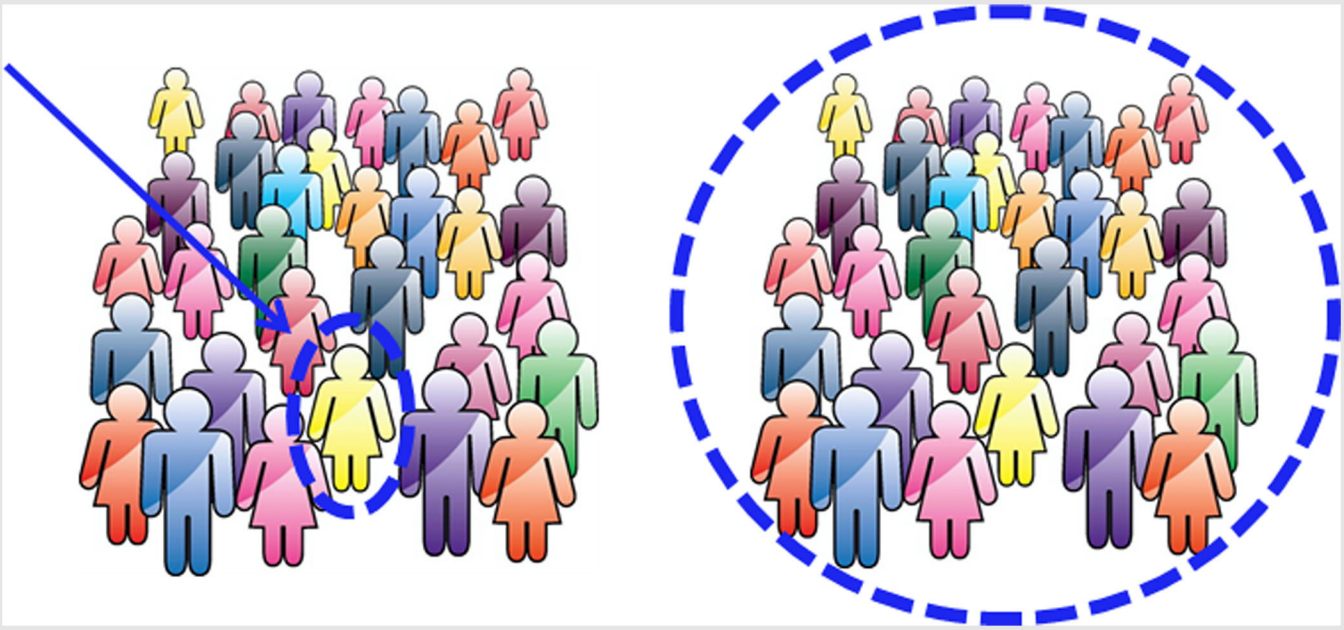
[3] Zawati MH, Parry D, Thorogood A, Nguyen MT, Boycott KM, Rosenblatt D, Knoppers BM. Reporting results from whole-genome and whole-exome sequencing in clinical practice: a proposal for Canada? J Med Genet. Published online September 27, 2013.

[4] Black L, McClellan KA, Avard D, Knoppers BM. Intrafamilial Disclosure of risk for hereditary breast and ovarian cancer: points to consider. J Community Genet. 2013;4:203–14.

4.1.1 Introduction to Genetic Testing and Screening

Genetic Testing

Genetic Screening



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Genetic testing and screening are two different things.

Genetic testing is offered to a specific individual. The purpose of genetic testing is to diagnose a genetic condition based on family history or symptoms.

Genetic screening is offered to populations who are at increased risk for genetic condition(s) but who do not demonstrate any symptoms. Genetic screening can either be offered to the entire population or to a targeted group. It is done to detect the presence of a condition before the onset of signs and symptoms to allow for intervention, including genetic counselling, that helps prevent manifestations of the condition if left undetected and untreated. It can also be done in order to identify an individual who is a carrier or a couple who are silent carriers of a recessive mutation that does not increase the risk to the individual or couple, but increases the risk of occurrence of the condition in question in their offspring. Screening is an initial step and involves additional tests for a definitive diagnosis.

4.2.1 Genetic Testing in Children

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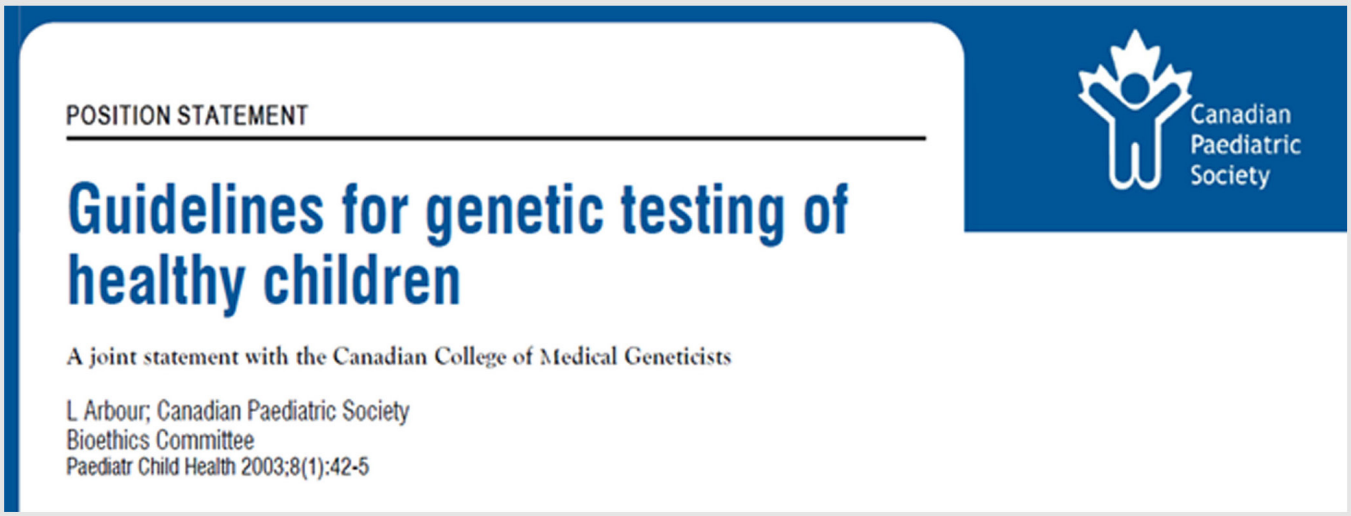
The most common type of genetic testing that is used for children is diagnostic testing or genetic testing to diagnose a childhood condition or conditions that can be treated medically during childhood.

There are many other genetic tests that are available to children. A number of them are not recommended. For example, genetic testing to identify conditions that do not become apparent until adulthood, such as some types of cancer, cardiovascular disease, and other single-gene conditions, are generally not recommended during childhood. Likewise, susceptibility or predictive testing to identify which people have a higher chance of getting a disease before symptoms appear are not recommended during childhood except possibly for hypertension, certain types of cardiac conditions, and certain hereditary cancers such as familial polyposis. Some tests should be used with caution in certain situations, for example, pharmacogenetic testing to predict the individual's response to a drug or course of therapy.

4.2.2 Diagnostic Genetic Testing in Children


POSITION STATEMENT

Guidelines for genetic testing of healthy children

A joint statement with the Canadian College of Medical Geneticists

L Arbour; Canadian Paediatric Society Bioethics Committee
Paediatr Child Health 2003;8(1):42-5

Canadian Paediatric Society

Diagnostic testing is genetic testing to diagnose a childhood condition or conditions that can be treated medically during childhood. An example is retinoblastoma (a cancerous tumour of the eye).

These tests are done in order to access treatment or management of a suspected genetic condition, where the treatment is likely to positively impact the child's condition. Genetic testing is offered if a child has symptoms and the test is used to confirm a medical diagnosis. There are also true predictive testing situations where genetic testing will enhance monitoring, treatment, or prevention in a healthy child at risk for a genetic condition but who does not have symptoms.[1]

The Canadian Paediatric Society (CPS) and the Canadian College of Medical Geneticists (CCMG) have developed guidelines for genetic testing of children.

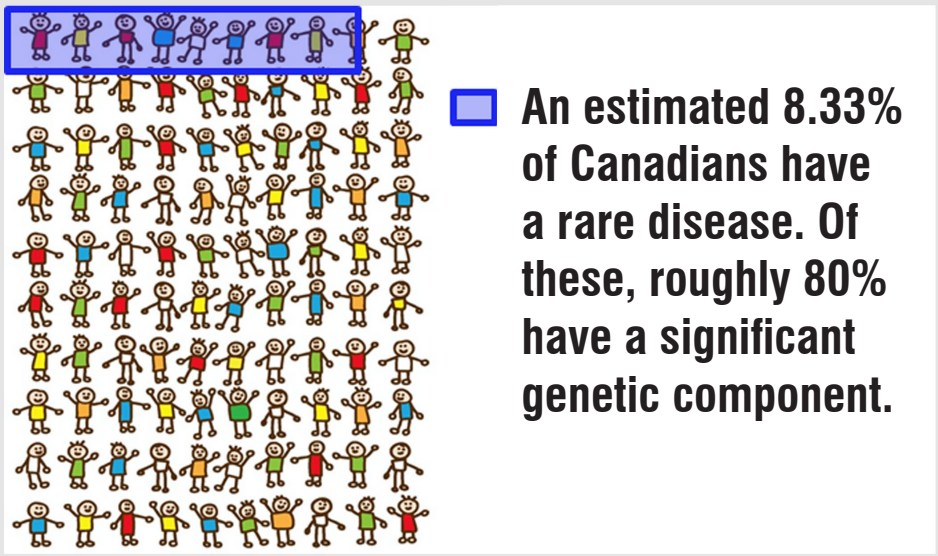
Highlights of the Guidelines for Genetic Testing of Healthy Children[1]

- The best interests of the child should be the primary consideration when contemplating testing.
- Parents should be informed of potential psychological and social risks associated with testing.
- There should always be appropriate counselling and genetic service involvement.
- Timely medical benefit to the child should guide genetic testing.
- When genetic conditions will not present until adulthood, testing should be delayed until the child is competent to decide whether they want the information.
- When carrier status for conditions is important only in reproductive decision-making, testing of children should be discouraged until the child is able to participate fully in the decision to be tested.
- Clinicians should consider requests for genetic testing by competent, well-informed adolescents for the purpose of reproductive decision-making.

For the full set of recommendations click here.

[1] Arbour L. Guidelines for genetic testing of healthy children. A joint statement with the Canadian College of Medical Geneticists Bioethics Committee, Canadian Paediatric Society (CPS) Ethics and Public Policy Committee, Canadian College of Medical Geneticists. Paediatrics & Child Health. 2003;8(1):42–5. Reference No. B03-01. Reaffirmed January 2011. Addendum April 2008.

4.2.3 Genetic Testing – Rare Diseases



Graphic created by CICH using a Shutterstock image and data from Canadian Organization for Rare Disorders www.raredisorders.ca

There are more than 7,000 single-gene diseases. Individually, each of these may be rare; however, when considered together, they are the cause of a significant number of childhood deaths, illnesses, and healthcare costs. Very often, rare disorders remain undiagnosed and have few therapies.

Approximately 30% of infants with a genetic disorder die before their first birthday.[1] Of the children who survive, many experience a comparatively high death rate over their lifetime.[2,3]

There are substantial costs to the healthcare system when caring for children with rare diseases. For example, approximately a third of childhood hospitalizations involve children with rare diseases.[4] These children also have a disproportionate number of hospital admissions and they tend to stay longer in hospital and incur larger hospital bills.

In 2013, the Canadian Institute of Health Research and Genome Canada funded "CARE for RARE" through its Personalized Medicine Initiative. A collaborative team from all regions of Canada, CARE for RARE is working to expand and improve the diagnosis and treatment of rare diseases.[5]

[1] Dodge JA, et al. The importance of rare diseases: from the gene to society. Arch Dis Child. 2011;96:791–2

[2] Dye DE, et al. The impact of single gene and chromosomal disorders on hospital admissions in an adult population. J Community Genet. 2011;2:81–90

[3] Yoon PW, et al. Contribution of birth defects and genetic diseases to pediatric hospitalizations. A population-based study. Arch Pediatr Adolesc Med. 1997;151:1096–103

[4] McCandless SE, Brunger JW, Cassidy SB. The burden of genetic disease on inpatient care in a children's hospital. Am J Hum Genetics. 2004;74(1):121–7

[5] Canadian Institute of Health Research and Genome Canada. CARE FOR RARE, http://care4rare.ca/about/overview/

Implications

The relatively small number of children with rare diseases in Canada, as well as in the world, presents challenges for rare-disease research. Recent governmental initiatives have been launched in an effort to support the application and integration of rare disease research. Orphanet Canada and the International Rare Disease Research Consortium work to rapidly spread research findings regarding rare diseases around the world and to facilitate action based on the findings.

4.3.1 Types of Genetic Screening

Preconception screening

Screening of women before they become pregnant

Prenatal screening

Screening of women during pregnancy

Newborn screening

Screening of infants immediately after birth

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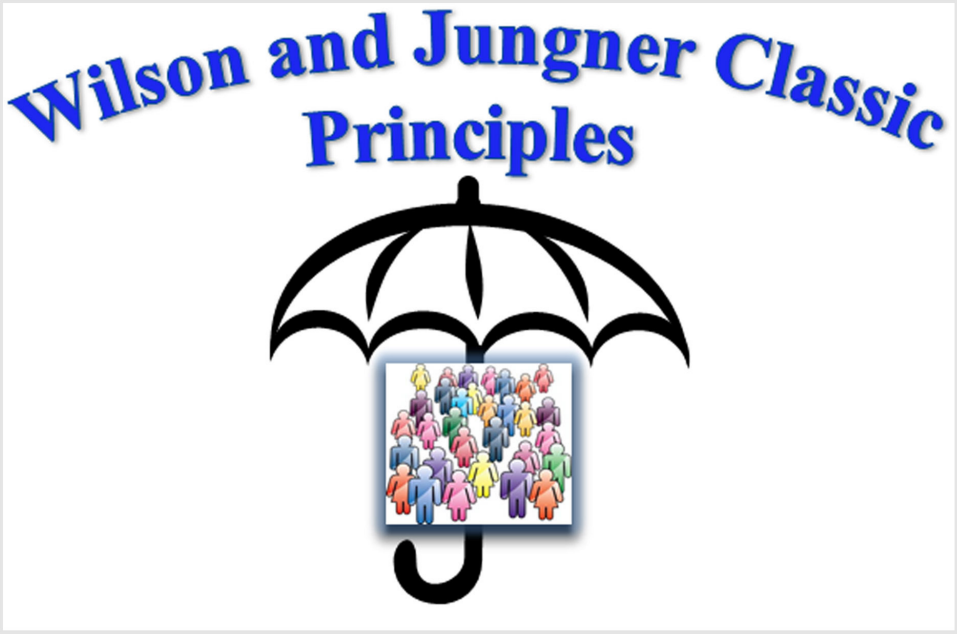
Screening may provide an early signal of a health problem in a child or adult who is not sick at the time of testing per se. It does not provide a definitive diagnosis, but rather suggests that he or she should receive further testing. There are different approaches to genetic screening depending on the population and the stage of life.

4.3.2 Genetic Screening: Overall Principles



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No matter what the type of genetic screening, certain core principles should be followed before a program is introduced.

Principles of Screening

- The condition sought should be an important health problem.
- There should be an accepted treatment for patients with recognized disease.
- Facilities for diagnosis and treatment should be available.
- There should be a recognizable latent or early symptomatic stage.
- There should be a suitable test or examination.
- The test should be acceptable to the population.
- The natural history of the condition, including development from latent to declared disease, should be adequately understood.
- There should be an agreed policy on whom to treat as patients.
- The cost of case finding (including diagnosis and treatment of patients diagnosed) should be economically balanced in relation to possible expenditure on medical care as a whole.
- Case finding should be a continuing process and not a "once and for all" project.

Source: Wilson JMG, Jungner G. Principles and practice of screening for disease. Geneva: World Health Organization; 1968.

Implications

In justifying population screening, it is important to provide benefits to the people who will be screened. Benefits require that screening be monitored and that appropriate clinical follow-up is provided based on the screening results. Early access to treatment and support greatly enhances alleviation of disease symptoms, reduces risk factors, and initiates observation measures for further disease signs and symptoms.

4.3.3 Preconception Screening

Image credit: Shutterstock

Preconception screening takes place prior to pregnancy. There are a number of situations where women might consider preconception screening. While the extent of preconception screening in Canada is unknown, the Society of Obstetrician and Gynaecologists of Canada provides guidelines for taking a preconception history for assessment and counselling.

Reasons for Preconception Screening

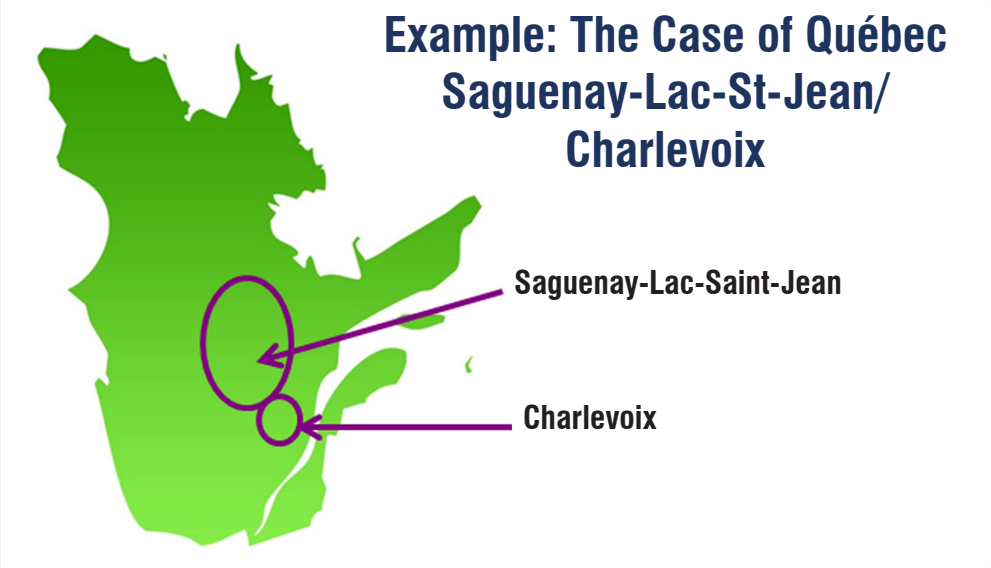
- Family history of an inherited disorder (e.g., a familial chromosome rearrangement).
- Diagnosis during in vitro fertilization prior to implantation.
- Diagnosis prior to artificial insemination.
- People from a specific ethnic group (e.g., thalassemia in people of Mediterranean and Southeast Asian descent or Tay-Sachs in Jewish persons of Eastern European descent).

Implications

The Society of Obstetricians and Gynaecologists of Canada warns that there is not enough evidence about preconception genetic risk assessment, screening, and testing to provide a conclusive guide for its use among healthcare providers.[1] The Society highlights that developing new tests may create new challenges, such as inviting genetic discrimination in applying for medical insurance.

[1] Wilson RD. Genetic Considerations for a Woman's Pre-conception Evaluation. SOGC Committee Opinion No. 253, January 2011. http://www.sogc.org/guidelines/documents/gui253CO1101E.pdf.

4.3.4 Preconception Screening Among Specific Populations



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Preconception screening aims to identify people who might be carriers of certain genetic traits. Some screening programs are conducted with specific ethnic groups with higher than normal chances for developing a particular condition.
Some Canadian provinces offer preconception screening for:

- Sickle-cell disease among individuals of African or Caribbean ancestry,
- Alpha or beta thalassemia for individuals of Mediterranean or Asian descent,
- Tay-Sachs disease, Familial Dysautonomia, Fanconi Anemia, and Canavan disease for individuals of Ashkenazi Jewish ancestry,
- Single gene conditions unique in targeted populations (e.g., screening of Hutterites for cystic fibrosis).

The Case of Quebec Saguenay-Lac-St-Jean/Charlevoix

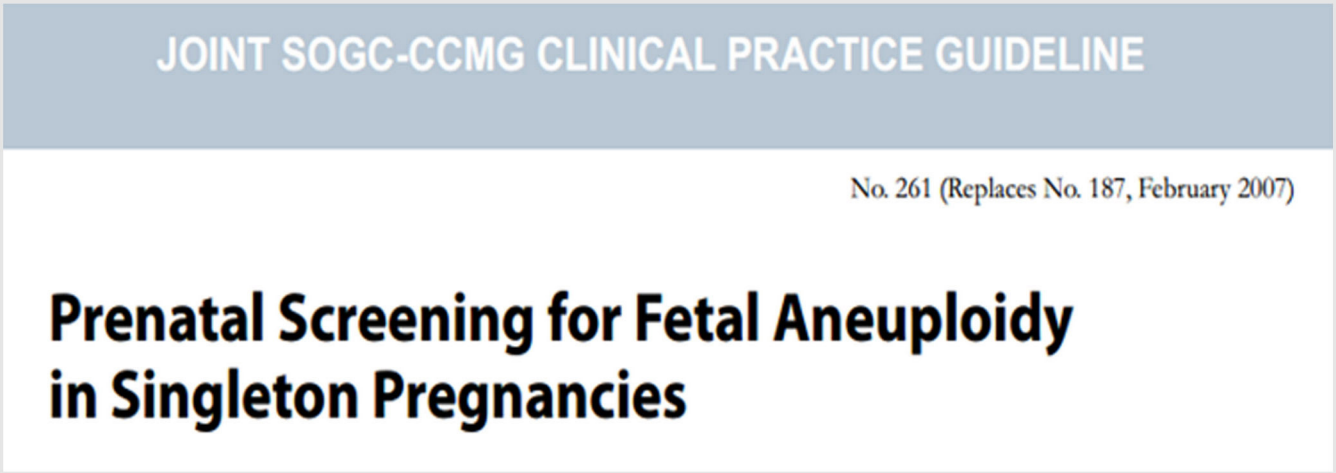
- A higher than expected number of people with ancestry from this region are carriers of one of four autosomal recessive genetic conditions: Autosomal recessive spastic ataxia of Charlevoix-Saguenay; Leigh syndrome, French-Canadian type; Tyrosinemia Type I; and Agenesis of the Corpus Callosum with Peripheral Neuropathy.
- If one or both parents have ancestors from this region, there is a higher likelihood of both being carriers and thus a higher risk of their child having one of these conditions.
- The region has a pilot-screening program that offers combined carrier testing for the four diseases (representing about 20% of the population) to any adult over 18 years of age who is known to have at least one grandparent originating from this region of Quebec and who is planning to have children.
- Screening is voluntary and must be initiated by the individual.
- Screening is preceded by an information session discussing benefits and possible disadvantages of screening.

Implications

Genetic screening programs require thoughtful planning, informed by engaging community stakeholders, to ensure benefit and to minimize any associated harms of genetic screening.

4.3.5 Prenatal Genetic Screening

The Society of Obstetricians and Gynaecologists of Canada and the Canadian College of Medical Geneticists have released prenatal screening guidelines.


JOINT SOGC-CCMG CLINICAL PRACTICE GUIDELINE

No. 261 (Replaces No. 187, February 2007)

Prenatal Screening for Fetal Aneuploidy in Singleton Pregnancies

Prenatal genetic screening is intended to provide information about the health of the fetus.

Prenatal screening can bring to light serious disabilities, such as congenital, genetic, and/or chromosomal problems. Generally, prenatal screening is offered as part of routine prenatal care, such as maternal serum screening, or if the mother is at risk of having a child with a serious genetic condition due to mature age or family history. See the following page for more information on prenatal blood screening programs in Canada.

Prenatal Screening Guidelines

The Society of Obstetricians and Gynaecologists of Canada and the Canadian College of Medical Geneticists recommend that screening for a condition should be undertaken only when the condition is considered to be serious enough to require intervention. They recommend that any screening program should:

- Be comprehensive and include information for parents and clinicians that is easily understood so that informed decisions can be made.
- Have timely access, a system to provide results and referral for follow-up testing, and access to treatment.
- Allow women and families to refuse testing at each step.
- Be evaluated.
- Have the ability to incorporate new technology.

Source: Chitayat D, Langlois S, Wilson RD. Prenatal Screening for Fetal Aneuploidy in Singleton Pregnancies. Joint SOGC-CCMG Clinical Practice Guideline No. 261 (Replaces No. 187, February 2007). http://www.sogc.org/guidelines/documents/gui261CPG1107E.pdf.

4.3.6 Prenatal Blood Screening Programs in Canada

Image credit: Shutterstock

Most provincial and territorial health insurance programs cover prenatal blood screening for chromosomal anomalies (Down syndrome and Trisomy 18) and neural tube defects. The programs usually include provincial coordination of education and evaluation, and participation is based on individual choice. It appears that participation is influenced by the preferences of both healthcare professionals and women. Individual provinces set their own policies regarding which prenatal screening blood tests are used for their program. This choice is partly determined by access to adequately equipped and staffed ultrasound facilities.

The Society of Obstetricians and Gynaecologists of Canada and the Canadian College of Medical Geneticists recommend that all pregnant women in Canada, no matter what age, should be offered the option of a prenatal screening blood test for the most common, clinically significant fetal chromosomal anomalies. They should also be offered a second trimester ultrasound for assessment of fetal anatomy, which should be done concurrently with counselling. The Society also provides minimum guidelines for the different testing methods that are available for use.[1]

[1] Chitayat D, Langlois S, Wilson RD. Prenatal Screening for Fetal Aneuploidy in Singleton Pregnancies. Joint SOGC-CCMG Clinical Practice Guideline No. 261 (Replaces No. 187, February 2007). http://www.sogc.org/guidelines/documents/gui261CPG1107E.pdf

Triple/Quadruple Screen

Triple/quadruple screen is the most common prenatal blood screening program offered in Canada. In this test, blood is taken from the pregnant woman to screen for a specific chromosomal anomaly in her fetus such as:

- Down syndrome, where a person has 47 chromosomes instead of the usual 46 – an extra chromosome 21.
- Trisomy 18, where a person has a third copy of chromosome 18, instead of the usual two.
- Neural tube defect, where there is an opening in the spinal cord or brain that occurs very early in development.

Implications

Prenatal screening results may cause mixed emotions for women and families, including stress, anxiety, relief, guilt, and questions about with whom to share the information. It is essential that women get professional genetic counselling to provide psychological support and to explain the results. The support should involve health professionals, including medical geneticists and genetic counsellors, who are knowledgeable about the challenges and limitations before, during, and after the screening. Families need this support since the decision to undergo prenatal testing is a personal choice based on values and experiences. Some families will want to know about genetic risk in order to prepare for caring for a child with a genetic condition, some may want the information to help them to decide whether to carry on with a pregnancy or terminate, while others may want the information to plan for future pregnancies.

4.3.7 Non-Invasive Prenatal Screening Methods

Image credit: Shutterstock

Following screening, genetic testing can occur during pregnancy for Down syndrome, Trisomy 18, and other conditions. Two major types of genetic testing are available: amniocentesis and chorionic villus sampling; however, these tests can increase the risk of miscarriage.

New non-invasive prenatal screening can now be conducted on a mother's blood samples. These tests analyze trace amounts of the baby's genetic information (DNA) that is present in the mother's blood. These tests are more reliable than the other tests, have a higher detection rate (e.g., 99% for Down syndrome), and can be carried out early in pregnancy (after 10 weeks).

The use of non-invasive prenatal screening varies from province to province. It is available privately (paid for by the patient) in British Columbia,[1] Manitoba, and Ontario[2] as an alternative to amniocentesis. Patients in Quebec can access these tests through private labs.

The Society of Obstetricians and Gynaecologists of Canada recommends using appropriate guidelines when offering prenatal non-invasive screening. The Society recommends that this type of screening should be an option after a positive result from currently used serum and ultrasound screening techniques for women wishing to avoid invasive testing. Early identification of a positive result may improve treatment and prognosis. The Society states that further studies are needed to determine if this method can be used reliably as a first screening approach in average-risk pregnancies.[3]

[1] BC Prenatal Genetic Screening Program, http://www.perinatalservicesbc.ca/ScreeningPrograms/PrenatalGeneticScreening/healthcare-providers/NonInvasivePrenatalTesting/default.htm

[2] Prenatal Screening Ontario. For Parents: Non-invasive Prenatal Testing (NIPT) Factsheet. 2012. http://www.mountsinai.on.ca/care/pdmg/NIPT%20info%20sheet%20for%20parents%2029_11_2012.pdf

[3] Langlois S, Brock J. Current status in non-invasive prenatal detection of down syndrome, trisomy 18, and trisomy 13 using cell-free DNA in maternal plasma. SOGC Committee Opinion No. 287, February 2013. J Obstet Gynaecol Can. 2013;35(2):177–81

Implications

Non-invasive prenatal screening technology requires further research. Genome Canada, Dr. François Rousseau at Université Laval, and Sylvie Langlois at University of British Columbia are among the leading interprofessional researchers based in eight Canadian universities and five European universities. They will compare different genetic technologies for their effectiveness in successfully detecting genetic conditions using the mother's blood.[4]

Non-invasive prenatal screening is currently not insured in Canada; however, it is available privately to those people who have the means to pay.

The decisions that families face regarding genetic screening are complex. If a fetus could inherit a serious health problem, the decisions facing the mother and family can be difficult. Access to appropriate counselling and support during these times is essential. To help women and families understand the implications, informed consent is an important component of the medical decision process.

[4] Rousseau F, Langlois S. Personalized genomics for prenatal aneuploidy screening using maternal blood (Pegasus). GénomeQuébec Inc. http://www.genomequebec.com/156-en/project/personalized-genomics-for-prenatal-aneuploidy-screening-using-maternal-blood-pegasus-.html

4.3.8 Newborn Genetic Screening

Image credit: Shutterstock

Newborn screening is a type of population screening to detect rare and serious conditions that can be easily and hastily treated before serious symptoms occur. Such screening tests can make the difference between healthy infant and child development, or lifelong disability, or even death for the child.

Because of their widespread and well-established success, newborn screening programs have been in existence for over 50 years in Canada and in most western countries. To date, newborn screening has traditionally been limited to diseases for which early detection and treatment offer direct medical benefits for the child. It is typically done by taking a sample of blood from the baby's heel 24 to 48 hours after birth and placing the droplet on a special filter paper.

4.3.9 Availability of Selected Common Newborn Screening Tests, Canada, 2013

	Cystic fibrosis	Congenital hypothyroidism	Sickle cell disease	Hearing	Transferase deficient galactosemia	Medium-chain acyl-CoA dehydrogenase	Phenylketo-nuria (PKU)
NL		A		B		A	A
PE		A		A		A	A
NS	C	A	C	A	C	A	A
NB		A		A		A	A
QC		A	C	B/C		A	A
ON	A	A	A	A	A	A	A
MB	A	A	B	B	A	A	A
SK	A	Req'd by law		B	A	A	Req'd by law
AB	A	A		B		A	A
BC	A	A	A	A	A	A	A
YK	A	A	A	B	A	A	A
NT	A	A		B		A	A
NU-Kitimeot	A	A				A	A
NU-Kivilliq		A		B	A	A	A
NU-Baffin		A		B		A	A

A = Universally offered but not required
B = Offered to select populations or by request
C = Testing required or offered universally but not yet implemented

Graphic reproduced by CICH using data from: Newborn Screening in Canada Status Report. Updated June 21, 2013. Canadian Organization for Rare Disorders; http://raredisorders.ca/documents/CanadaJune21.pdf.

In Canada, every province/territory has a newborn screening program. The inherited diseases included in the provincial/territorial programs vary significantly as there are no nationwide standards for the conditions required for screening. All provinces screen for phenylketonuria (PKU), congenital hyperthyroidism (CH), and medium-chain acyl-CoA dehydrogenase deficiency (MCADD). Saskatchewan is the only province/territory that requires screening for CH and PKU by law. A number of provinces/territories also screen for conditions such as cystic fibrosis, and other metabolic and endocrine conditions.

In recent years, a number of new tests have been added. Some provinces have expanded the number of conditions required for screening, while others have not. The Canadian Organization for Rare Disorders provides the full list of conditions in newborn screening programs by province and territory.

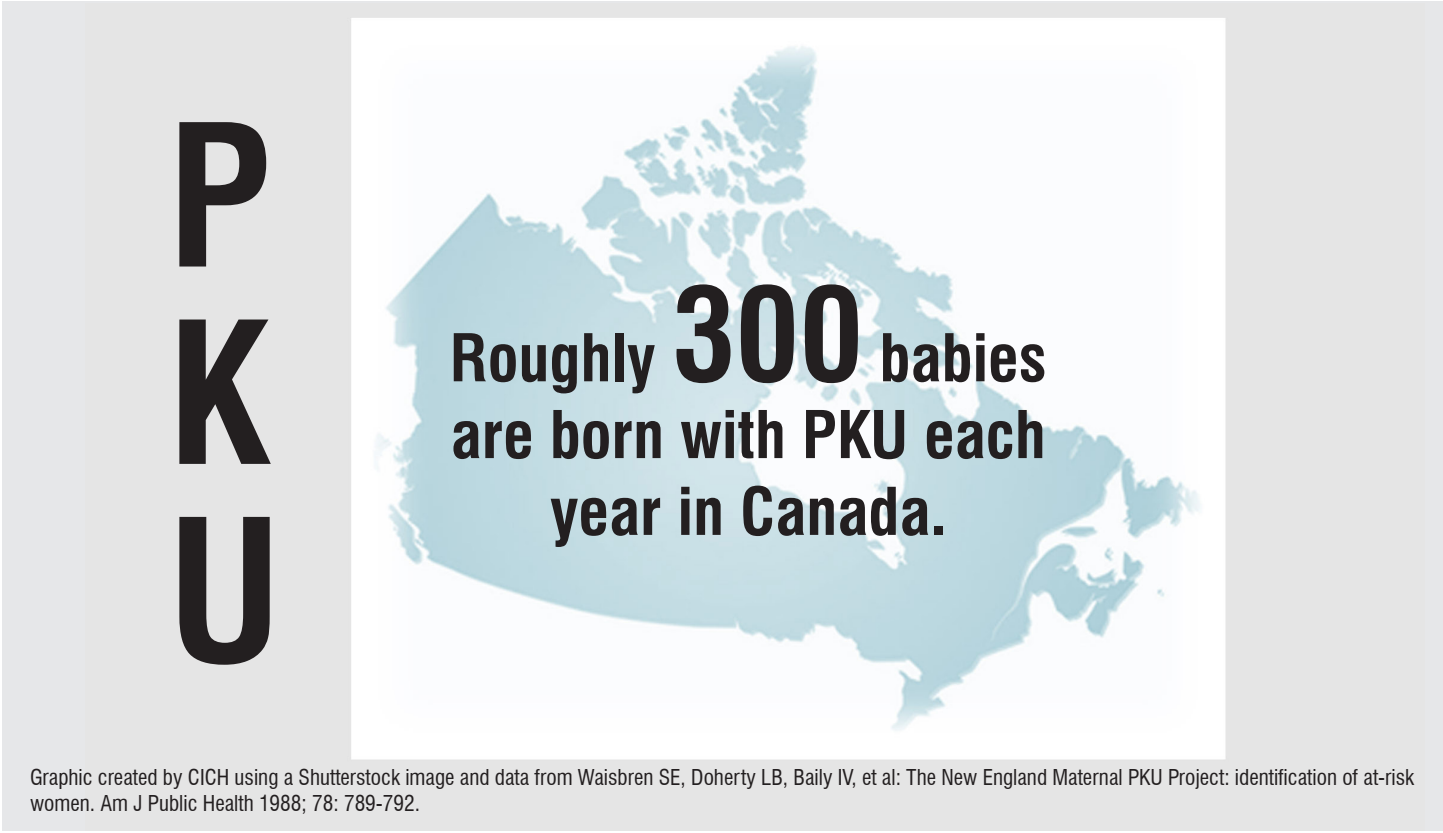
Consent for newborn screening is usually "implied," but parents have the right to opt out of newborn screening. Most provinces provide guidelines for formal documentation when the parents opt out of newborn screening.

Implications

In Canada, newborn screening programs are used for conditions that respond to early treatment and support, which alters the course or severity of the condition to improve the child's wellbeing. However, management or treatment for these conditions is a lifelong process that can be challenging and costly. Many provinces instituted newborn screening using tandem mass spectrometry, which screens for approximately 40 hereditary metabolic conditions consistent with the guidelines of the American College of Medical Genetics.[1] More consistent newborn screening policies need to be developed because the full range of newborn screening tests are not uniformly available across Canada.

[1] Green RC, Berg JS, Grody WW, Kalia SS, Korf BR, Martin CL, et al. ACMG Recommendations for Reporting of Incidental Findings in Clinical Exome and Genome Sequencing. Bathesda, MD : American College of Medical Genetics and Genomics; 2013. http://www.acmg.net/docs/ACMG_Releases_Highly-Anticipated_Recommendations_on_Incidental_Findings_in_Clinical_Exome_and_Genome_Sequencing.pdf

4.3.10 Newborn Genetic Screening – Specific Conditions – Phenylketonuria (PKU)



Graphic created by CICH using a Shutterstock image and data from Waisbren SE, Doherty LB, Baily IV, et al: The New England Maternal PKU Project: identification of at-risk women. Am J Public Health 1988; 78: 789-792.

Phenylketonuria (PKU) is a rare genetic condition in which a baby cannot "metabolize," or digest, an essential amino acid called phenylalanine that is found in foods with protein. If untreated, severe brain damage can result from the elevated levels of phenylalanine. PKU is a genetically inherited condition. Both parents must carry a mutation in the gene that is responsible for providing instructions on making an enzyme called phenylalanine hydroxylase. A baby with PKU must receive two copies of the gene mutation, one from each parent.[1] PKU is rare, with approximately 1 in 12,000 newborns in North America diagnosed, which translates to 300 babies born with PKU in Canada each year.[2]

[1] A.D.A.M Medical Encyclopedia, US National Library of Medicine. PubMed Health. http://www.ncbi.nlm.nih.gov/pubmedhealth/PMH0002150/ Retrieved June 1, 2012. [2] Waisbren SE, Doherty LB, Baily IV, et al. The New England Maternal PKU Project: identification of at-risk women. Am J Public Health. 1988;78:789–92.

Implications

Brain damage and developmental delay can only be prevented when babies are diagnosed with PKU early – that is, as soon as possible after birth, generally between 7 and 10 days – and treatment is started immediately. The most effective treatment is a special diet very low in the amino acid phenylalanine, which must be followed throughout life. Infants and children on the PKU diet require special infant formula and specialty low protein products.

Other treatment options, which involve special drugs that lower phenylalanine levels, are being researched and could potentially improve quality of life for people with PKU. When young women with PKU reach their reproductive years, it is essential that they are aware of their diagnosis. They must follow a strict diet before and during pregnancy to ensure that their baby is healthy.

4.3.11 Newborn Genetic Screening - Specific Conditions - Access to Treatment for (PKU), Canada, 2012

Province	Formulas	Low-Protein Foods	Shipping (to patient)
NL	✓ only 2 formulas	✓ only staples - pasta, bread mix, pizza shells, cheese	✓
PE	✓		
NS	✓	✓ only staples - baking mix, pasta, cracker toasts, rusks	✓ if distant from clinic
NB	✓	✓ only staples - bread mix, flour, pasta	✓
QC	✓	✓ up to $1,500/yr for Cambrooke Foods	✓ if distant from CLSC clinics
ON	✓	✓	✓
MB	✓	✓ up to $120/mo age 0-12; up to $250/mo age 13-18	✓ if distant from clinic
SK	✓	✓	✓ if distant from clinic
AB	✓	✓	
BC	✓		✓ if outside Lower Mainland

Graphic adapted by CICH from: Canadian PKU and Allied Disorders. http://canpku.org/images/pdf/coverage-pku-2012.pdf, Retrieved April, 2012

The cost of PKU infant formula is covered under provincial health insurance in all provinces for everyone with a provincial health card. However, other aspects of treatment, such as specialty foods, are not universally covered across Canada, nor are shipping costs for formula/food. The specialty foods are substantially more expensive than regular food.

4.3.12 Newborn Genetic Screening – Specific Conditions – Cystic Fibrosis

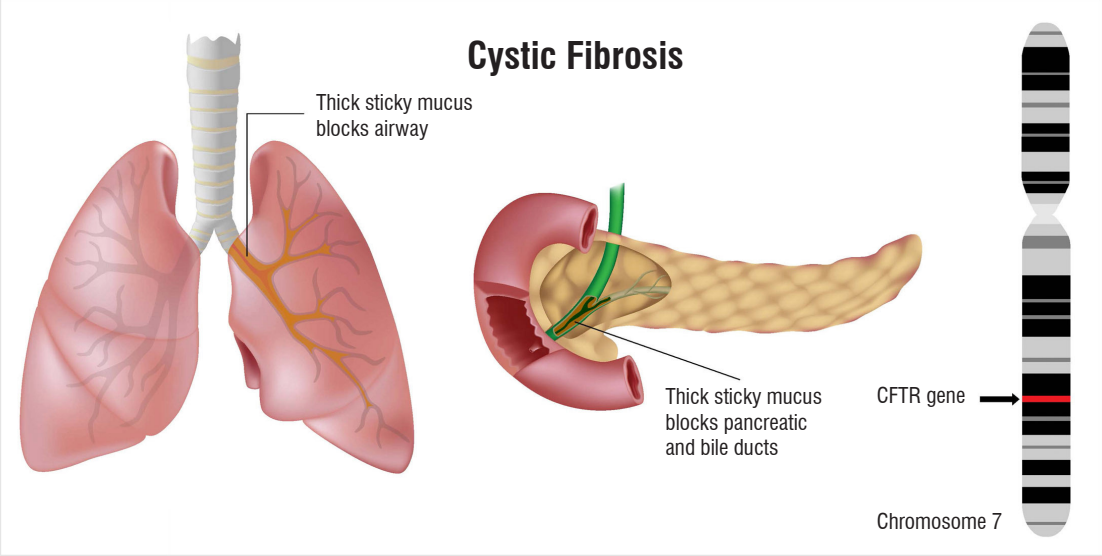


Image credit: Shutterstock

Cystic fibrosis is an inherited genetic condition. Both parents must carry a mutation in one of the two copies of the gene that causes cystic fibrosis (the CFTR gene) and each parent must pass the mutation on to their child. If a child receives two copies of the faulty gene – one from each parent – he or she will develop cystic fibrosis.

Cystic fibrosis is a serious illness that severely affects day-to-day living. Not all people are affected in the same way; however, common symptoms include thick mucus build up in the lungs and difficulties breathing. Mucus and protein also accumulate in the digestive tract, making it difficult to digest and absorb nutrients from food. Though cystic fibrosis affects both children and adults, most are diagnosed as infants.

Treatment consists of respiratory and nutritional therapy to help the body absorb foods and prevent blockages from the thick mucus. Newborns diagnosed with cystic fibrosis need specialist care and are normally followed by interprofessional cystic fibrosis teams in paediatric hospitals across Canada.[1]

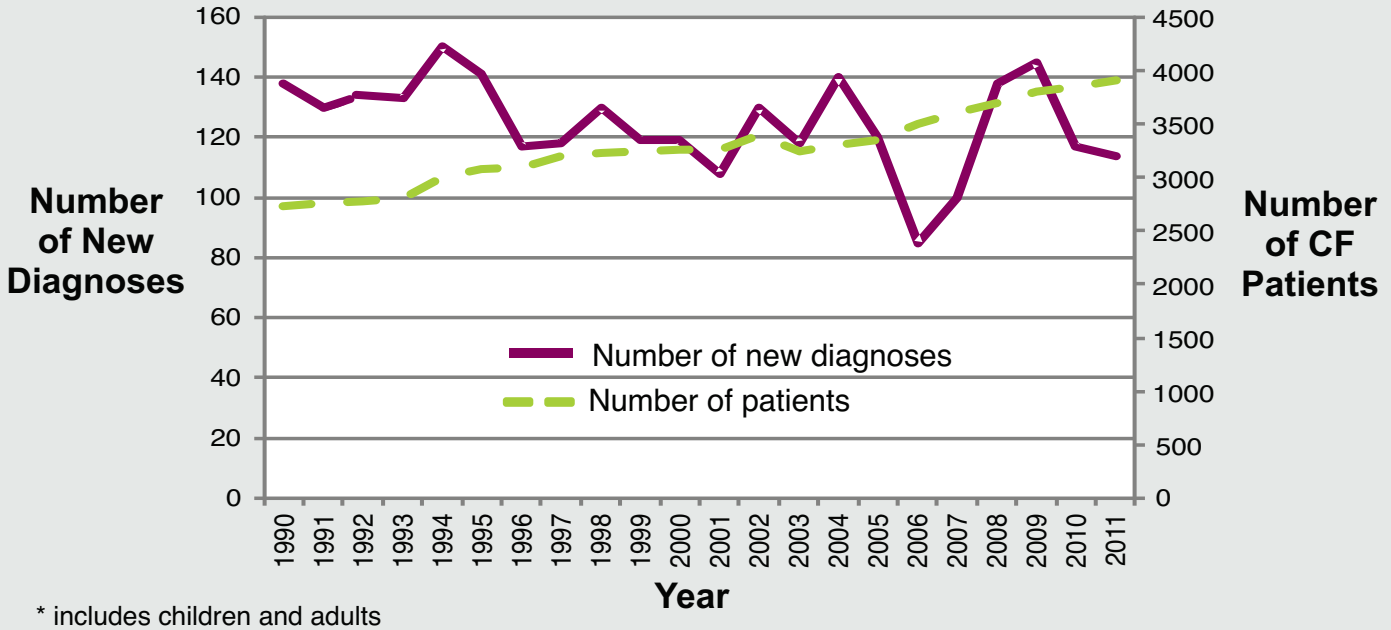
[1] Cystic Fibrosis Canada, About CF

Some Facts About Cystic Fibrosis

- One in every 3,600 children born in Canada has cystic fibrosis.
- 50% are diagnosed by 6 months of age, and 73% by the age of 2 years.
- The median age of people living with cystic fibrosis is 20 years. About 43% of people living with cystic fibrosis are 18 or younger.
- In 2011 there were 114 people diagnosed with cystic fibrosis.
- Children with cystic fibrosis used to die very young. Now, on average, people with this disorder live into their thirties and forties.
- Newborn screening for cystic fibrosis can help to improve the outcomes for those diagnosed with the condition, including reduced hospitalizations and a longer life expectancy.

Source: Cystic Fibrosis Canada. The Canadian Cystic Fibrosis Registry. 2011 Annual Report. http://www.cysticfibrosis.ca/cf-care/cf-registry/

4.3.13 Newborn Genetic Screening - Number of Cystic Fibrosis Patients* Seen in Clinics and Number of New Diagnoses, Canada, 1990 to 2011

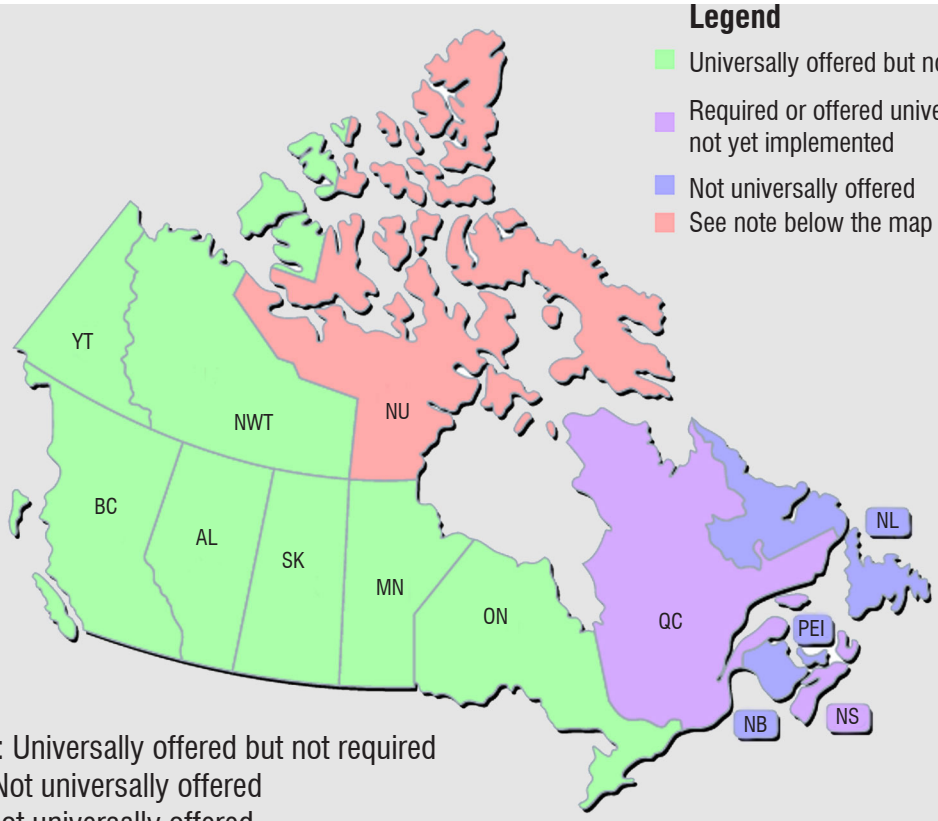


* includes children and adults

Graphic created by CICH using data from Canadian Cystic Fibrosis Patient Data Registry Report 2010 and the Canadian Cystic Fibrosis Registry 2011 Annual Report. Cystic Fibrosis Canada. www.cysticfibrosis.ca

In 2011, 114 Canadians were newly diagnosed with cystic fibrosis. This number decreased between 2004 and 2006 but increased again between 2007 and 2009. The increase may have been due in part to the introduction of newborn screening programs in several provinces. In 2011, 3,913 individuals with cystic fibrosis were seen in specialty clinics in Canada, and of those 1,675 were younger than 18 years of age.

4.3.14 Specific Conditions – Newborn Screening for Cystic Fibrosis



Note:

Nunavut regions:

- Kitimeot region: Universally offered but not required
- Kivilliq region: Not universally offered
- Baffin region: Not universally offered

Source: Newborn Screening in Canada Status Report. Updated June 21, 2013. Canadian Organization for Rare Disorders. http://raredisorders.ca/documents/CanadaJune21.pdf

Eight provinces/territories offer newborn screening for cystic fibrosis.

Early diagnosis and early treatment of cystic fibrosis can reduce hospitalizations and improve the quality of life and life expectancy of cystic fibrosis patients. Without newborn screening, most people are not diagnosed until they present with symptoms. By that time, early damage to the lungs and digestive system may be difficult to reverse. Research demonstrates that a newborn diagnosed early with cystic fibrosis will have an improved height, weight, nutritional status, lung function, and cognitive ability.[1,2,3]

[1] Newborn Screening for Cystic Fibrosis. Cystic Fibrosis Canada. http://www.cysticfibrosis.ca/?lang=en

[2] Farrell PM, Kosorok MR, Rock MJ, Laxova A, Zeng L, Lai HC, Hoffman G, et al. Early diagnosis of cystic fibrosis through neonatal screening prevents severe malnutrition and improves long-term growth. Wisconsin Cystic Fibrosis Neonatal Screening Study Group. Pediatrics. 2001;107(1):1–13

[3] Southern KW, Merelle MM, Dankert-Roelse JE, Nagelkerke AF. Newborn screening for cystic fibrosis. Cochrane Database Syst Rev. 2009;(1):CD001402

Implications

Newborn screening for cystic fibrosis raises a number of ethical questions. Because it can ultimately identify a child who is just a carrier but not affected, there are pros and cons to communicating this information to parents. If parents discover that their child is a carrier, they will have numerous questions about the long-term implications for their child and about their own health. This can be very stressful.

4.3.15 Newborn Genetic Screening - Specific Conditions - Sickle Cell Disease

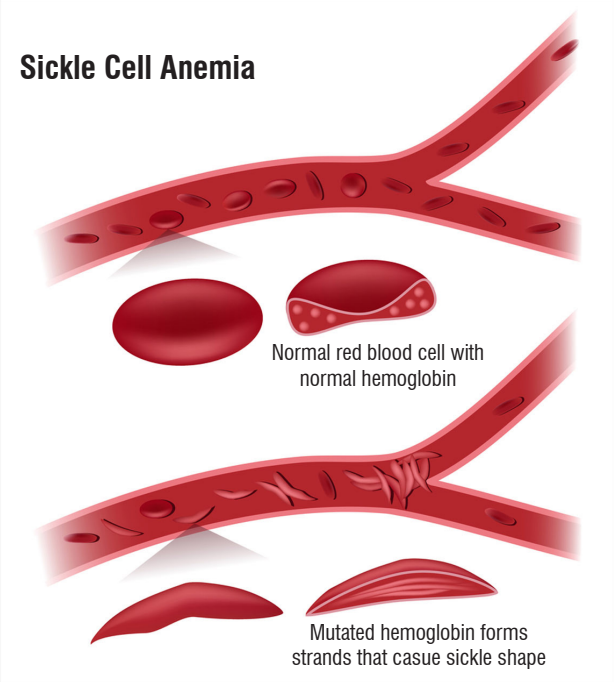


Image credit: Shutterstock

The signs and symptoms of sickle cell disease vary. Some people suffer mild symptoms, while others develop very severe symptoms and are often hospitalized for treatment. Sickle cell disease is present at birth, but many infants do not have symptoms until after four months of age.

Sickle cell disease is a genetic condition and is therefore passed from parent to child. It can have severe physical, psychological, and social consequences for newly diagnosed patients and their families. Some children will be relatively healthy. Others, however, are admitted to hospital for immediate care. Recognizing sickle cell disease early is the key to preventing complications.

Different treatments and medications can help to relieve symptoms that might occur with the illness. Most of these are linked to anemia, infection, and pain.[1] Others are associated with disease complications.

[1] Sickle Cell Disease Association of Canada. http://www.sicklecelldisease.ca/

Sickle cell disease is a condition that affects the hemoglobin contained in a child's red blood cells. It is often diagnosed during childhood but can also be detected during newborn screening.

For information on the sickle cell disease screening programs offered throughout Canada, click here.

For more information about Sickle Cell Disease see The Montreal Children's Hospital and the McGill University Health Centre's Sickle Cell Disease Family Handbook

4.3.16 Newborn Genetic Screening - Specific Conditions - Some Facts About Sickle Cell Disease

...the number of people in Canada living with Sickle Cell Disease.

1 in 2,500 Canadian babies are born with Sickle Cell Disease.

That is, between 100 and 150 babies born each year in Canada will be diagnosed.

Graphic created by CICH using images from Shutterstock and data from Sickle Cell Disease Association of Canada. http://www.sicklecelldisease.ca/education/general-knowledge/ and Shutterstock images.

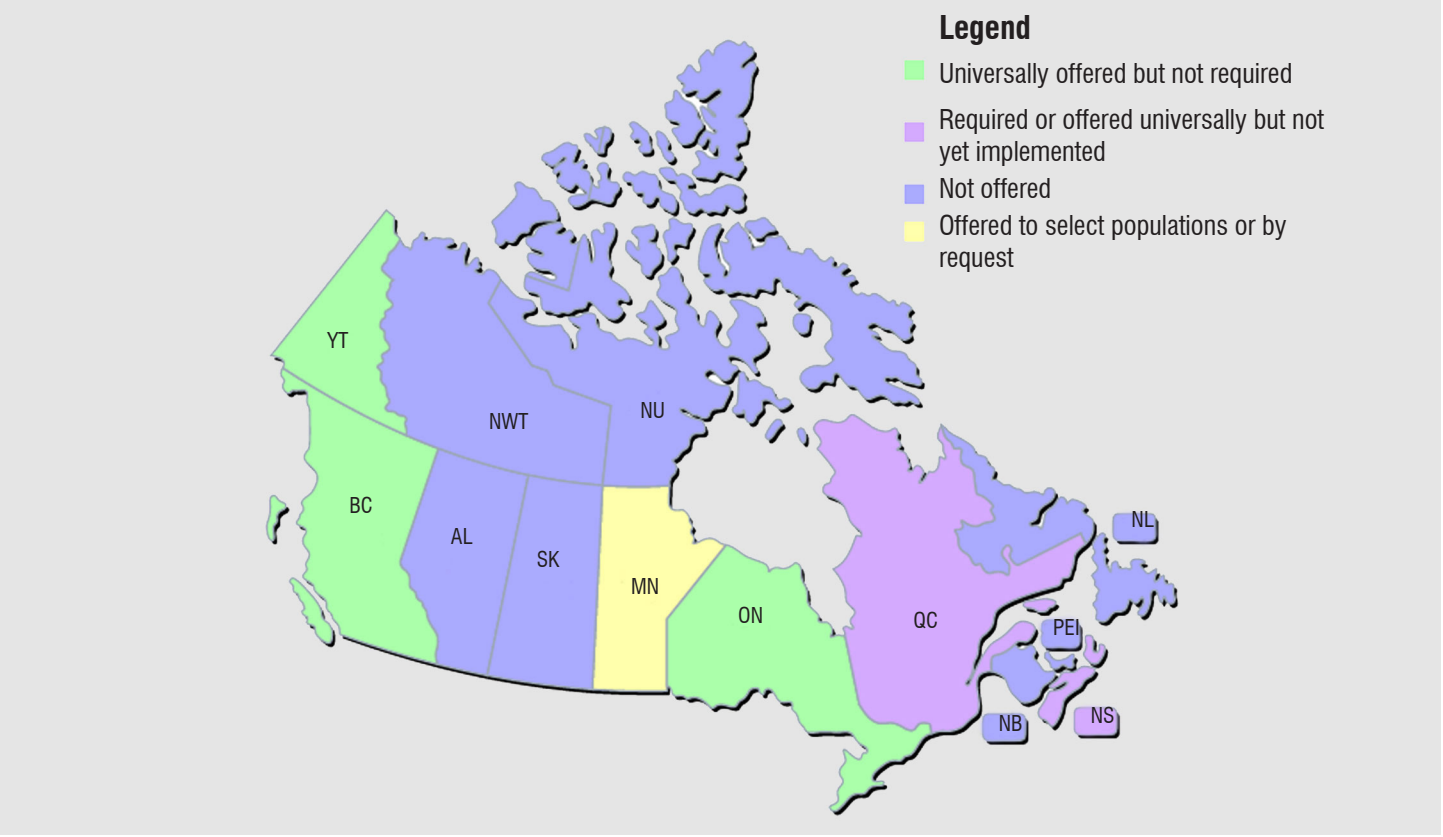
Sickle cell disease affects millions of people around the world. In Canada, there are about 2,000 people living with the disease. Up to 1 in every 2,500 babies born in Canada will have the disease.[1] Sickle cell disease is most common in families of African ancestry, but children of Middle Eastern, Mediterranean, sub-Saharan African, and Asian ancestry are also affected.

The Canadian Paediatric Society recommends that all children and youth new to Canada who travel from regions where sickle cell disease is common and do not have reliable, previous documentation should be screened for sickle cell disease.[2]

1 Sickle Cell Disease Association of Canada. http://www.sicklecelldisease.ca/education/general-knowledge/

2 Canadian Paediatric Society, Caring for Kids New to Canada. http://www.kidsnewtocanada.ca/conditions/sickle-cell

4.3.17 Specific Conditions – Newborn Screening for Sickle Cell Disease



Ontario, British Columbia, and the Yukon are the only provinces/territories in Canada that have universal newborn screening programs for sickle cell disease. Manitoba offers screening for select populations upon request. In Nova Scotia and Quebec, testing is required or offered universally but not yet implemented.[1]

[1] Canadian Organization for Rare Disorders. Newborn Screening in Canada Status Report. June 2013. http://www.raredisorders.ca/documents/CanadaJune21.pdf

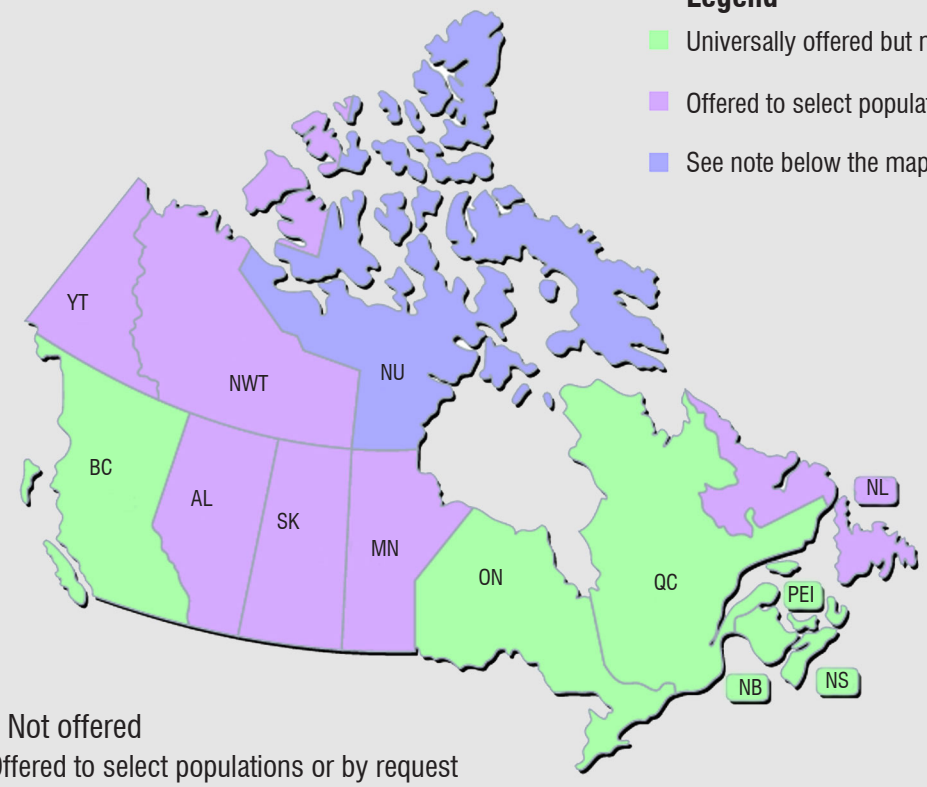
Implications

The United Nations and the World Health Organization recognize that sickle-cell disease is one of the world's foremost genetic diseases. As a result, the General Assembly of the United Nations asked member states, including Canada, to support global efforts to address sickle cell disease. Efforts include public health programs for newborn screening and basic research on the disease.[2] In Canada, there has been a call for a National Strategy for Sickle Cell Disease and Thalassemic Disorders. This bill was introduced in the House of Commons in 2011 but has not progressed further than first reading.[3] One of the critical issues related to sickle cell disease screening is that it identifies carriers. This raises dilemmas for parents, has minimal benefit for the child, and, if poorly understood, may impact the child's psychosocial wellbeing.

[2] United Nations. General Assembly. Sixty-third session. Agenda item 155. Resolution adapted by the General Assembly. 2009. http://www.worldlii.org/int/other/UNGARsn/2008/277.pdf.

[3] House of Commons of Canada. Bill C-221. An Act respecting a Comprehensive National Strategy for Sickle Cell Disease and Thalassemic Disorders. http://www.parl.gc.ca/HousePublications/Publication.aspx?DocId=5092338&Language=E&Mode=1&File=4

4.3.18 Specific Conditions – Newborn Hearing Screening



Note:

Nunavut regions:

- Kitimeot region: Not offered
- Kivilliq region: Offered to select populations or by request
- Baffin region: Offered to select populations or by request

In Canada, about one to three newborns for every 1,000 born will have a permanent hearing loss. Each year, between 380 and 1,200 newborn babies are newly diagnosed with severe hearing loss. In about 50% of these cases, genetics is to blame. If hearing loss is diagnosed in newborns, they have the opportunity of early treatment. This results in better outcomes for children when compared to those who are diagnosed at a later age.[1]

The Canadian Pediatric Society recommends universal hearing screening for all newborn babies in Canada. Newborn screening varies across the country. Currently five provinces offer universal newborn hearing screening; the others offer screening to targeted at-risk populations.

[1] Patel H, Feldman M. Universal newborn hearing screening. Canadian Paediatric Society position statement. Canadian Paediatric Society, Community Paediatrics Committee. Paediatr Child Health. 2011;16(5):301–5

5.1.1 What Are Congential Anomalies?

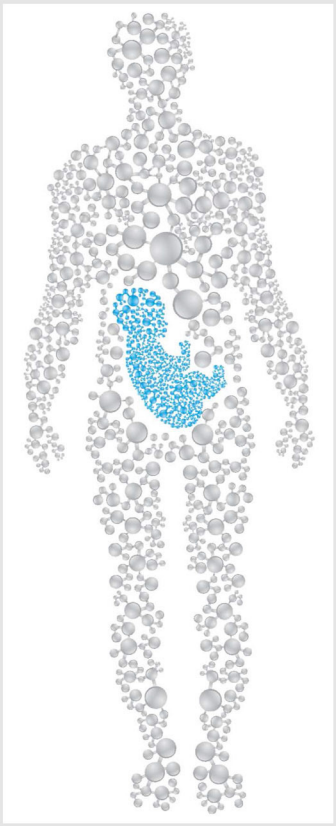


Image credit: Shutterstock

Congenital anomalies, sometimes called birth defects, happen during prenatal development. They include abnormalities of structure, function, or metabolism. They are present at birth but may not be diagnosed until later in life. They can result in physical or mental disability, affect a child's development, and, in severe cases, can be fatal.[1]

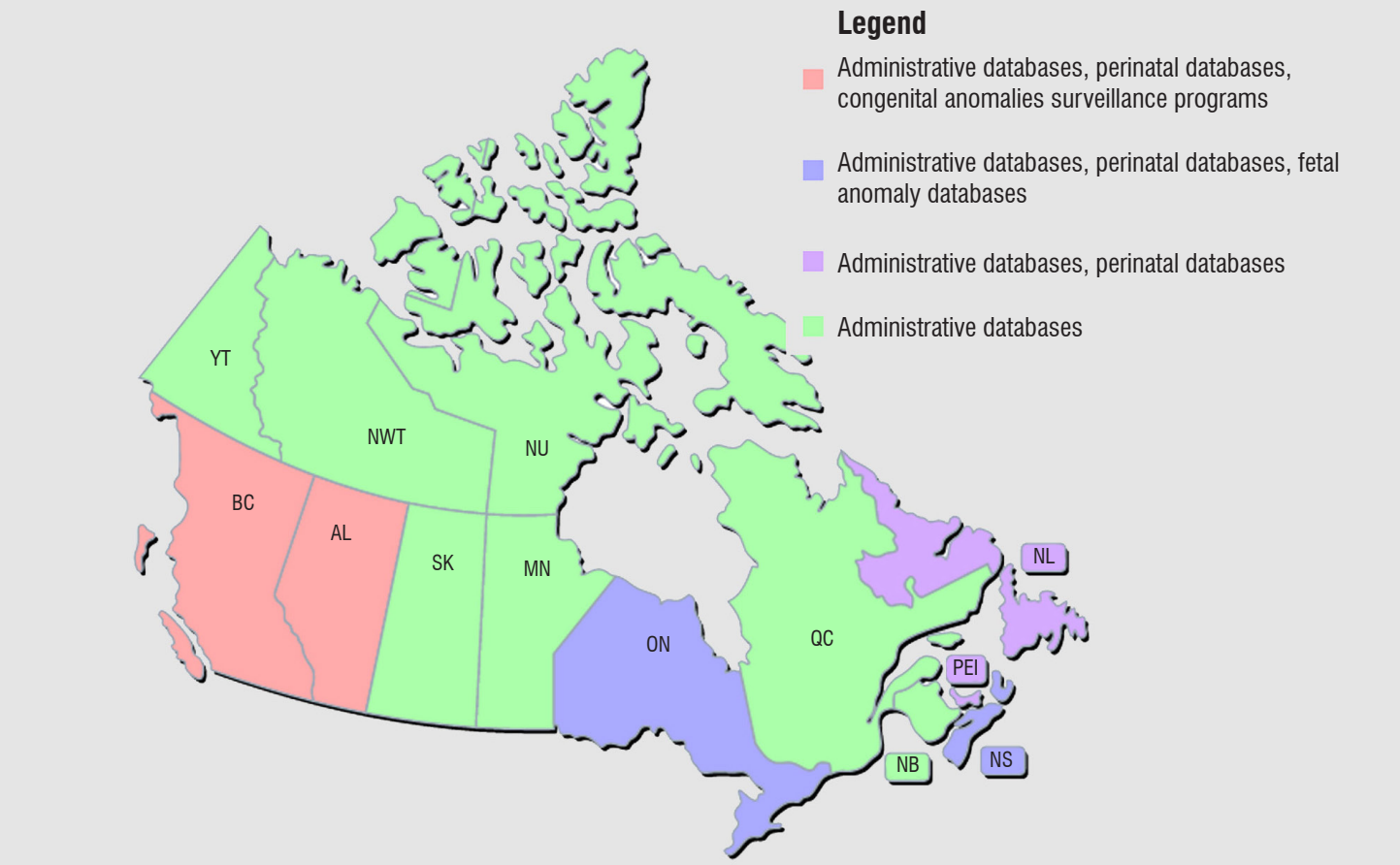
Congenital anomalies are a leading cause of death among foetuses and infants[2] and can greatly influence quality of life. They are costly to both families and the healthcare system.[3]

[1] Definition adapted from Health Canada. Congenital Anomalies in Canada — A Perinatal Health Report, 2002. Ottawa: Minister of Public Works and Government Services Canada, 2002.

[2] Lowry R, Sibbald B, Bedard T. Alberta Congenital Anomalies Surveillance System Eighth Report 1980–2007. Government of Alberta Report, 1-45. Canadian Perinatal Health Report 2008 Edition (pp. 317). Ottawa: The Public Health Agency of Canada; 2009.

[3] Public Health Agency of Canada and the Congenital Anomalies Surveillance Network. Towards Enhanced Congenital Anomalies Surveillance in Canada (pp. 1–14). Ottawa; 2008.

5.1.2 Monitoring Congenital Anomalies



The Canadian Congenital Anomalies Surveillance System (CCASS) gathers and collates data from hospital databases and from some provincial congenital anomalies surveillance systems. However, the collection and recording of information regarding congenital anomalies is not standardized across the country.1 Different provinces test for different conditions, use different sources of data, and assess the presence of congenital anomalies in their populations differently. For example, some include data on fetal anomalies from pregnancies terminated following a prenatal diagnosis, while others do not.[1,2]

[1] Little J, Potter B, Allanson J, Caulfield T, Carroll JC, Wilson B. Canada: Public Health Genomics. Public Health Genomics. 2009;12:112–120.

[2] Demographics and Risk Indicators Working Group, C.C.A.S.N., Public Health Agency of Canada. Congenital Anomalies Surveillance in Canada: Results of a 2006–2007 Survey on Availability of Selected Data Variables in Canadian Provinces and Territories. Ottawa: Public Health Agency of Canada, 2010

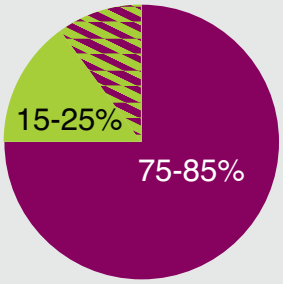
Implications

The lack of standardization makes it difficult to compare data across provinces and territories. A new initiative to improve the current system by strengthening surveillance at the provincial and territorial level is underway to address the limitations in collection and reporting of data in the future. Congenital anomalies are an important cause of childhood death, chronic illness, and disability. There is a need to develop standardized methods of coding data relating to congenital anomalies, along with better registration and surveillance. Further research about the causes of congenital anomalies is needed.

5.1.3 Causes of Congenital Anomalies

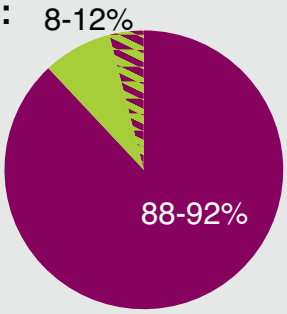
Recognized genetic conditions:
Chromosome and single gene causes

15% to 25% are due to recognized genetic conditions



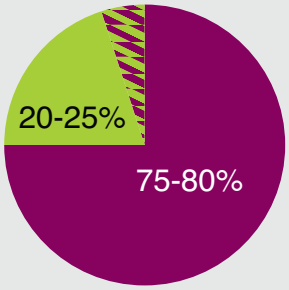
Environmental factors:
Maternal-related conditions, drug or chemical exposures

8% to 12% are due to environmental factors



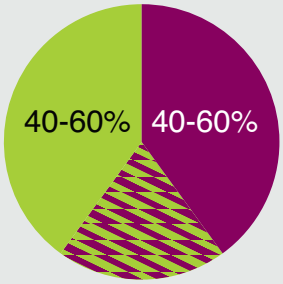
Multifactorial inheritance*

20% to 25% are due to multifactorial inheritance



Unexplained causes

40% to 60% are due to unexplained causes

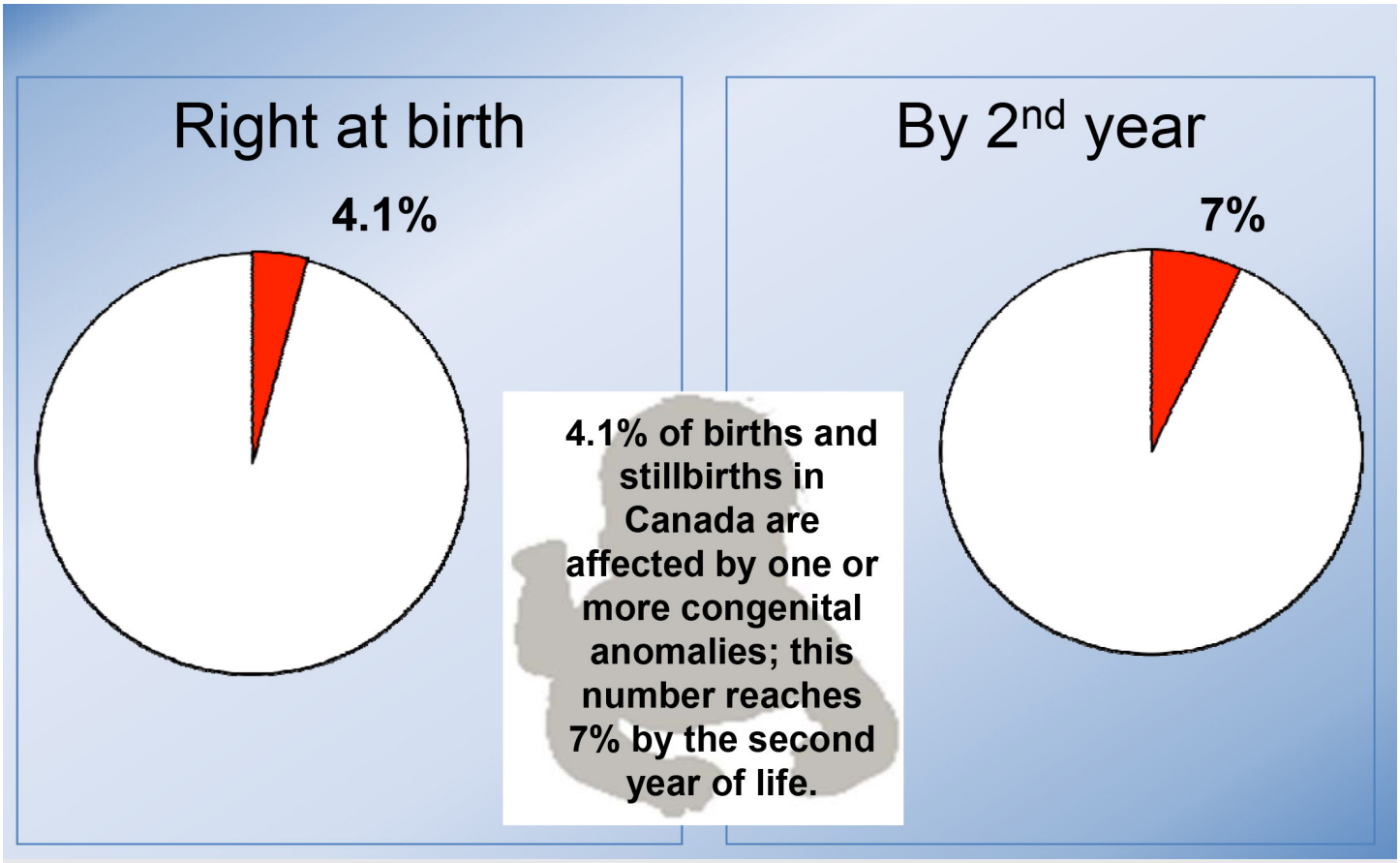


* A congenital anomaly is considered to be multifactorial (or polygenic) in origin when there is a combined influence of (a number of) genes and environmental factors that interfere with normal embryologic development. Multifactorial inheritance is considered when there appears to be a genetic component but there is no clear Mendelian pattern of inheritance. Multifactorial inheritance is the underlying etiology of most of the common congenital anomalies.

Graphic created by CICH using data from Health Canada. Congenital Anomalies in Canada — A Perinatal Health Report, 2002. Ottawa: Minister of Public Works and Government Services Canada, 2002.

Some congenital anomalies are caused by mutations in a single gene or damage to a specific chromosome. Other congenital anomalies are due to exposure to environmental hazards or drugs during pregnancy. Others result from a combination of genetic and environmental influences. However, the cause of most congenital anomalies is unknown. Most children with congenital anomalies are born to mothers with no family history and no known risk factors.

5.1.4 Prevalence of Congenital Anomalies in Canada



Graphic created using a Shutterstock images and data from Health Canada. Congenital Anomalies in Canada — A Perinatal Health Report, 2002. Ottawa: Minister of Public Works and Government Services Canada, 2002.

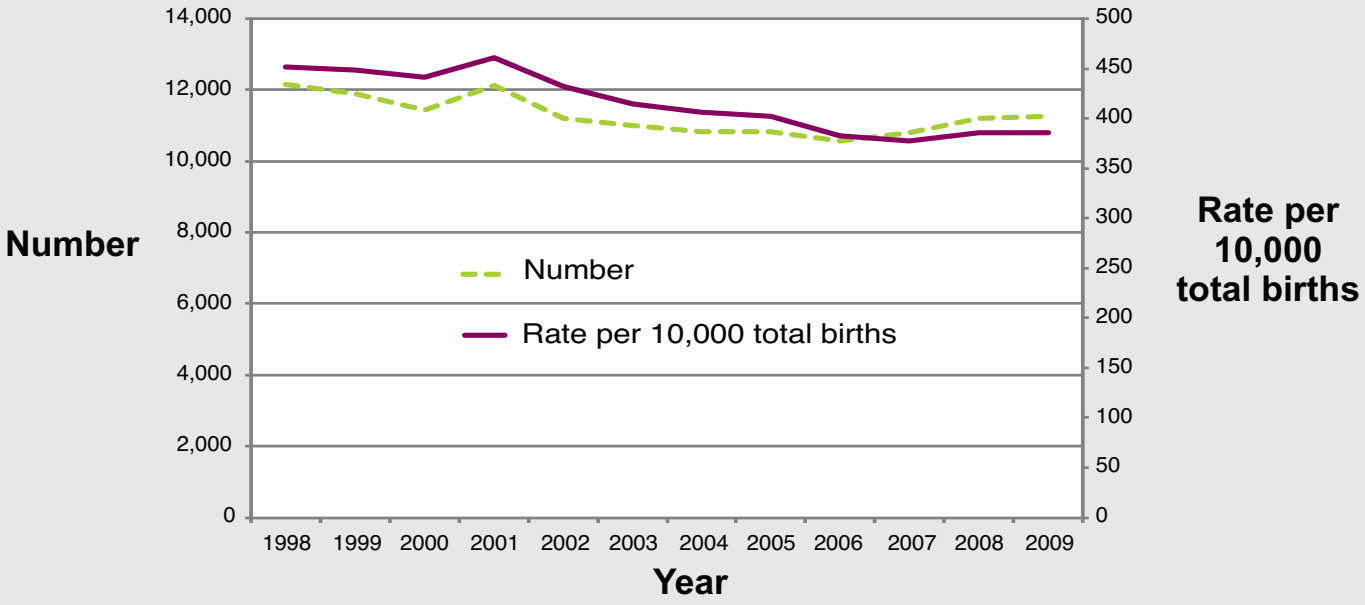
In 2007, 4.1% of all births in Canada, including stillbirths, were affected by one or more congenital anomalies. It is estimated that major congenital anomalies affect an estimated 3% of all births.[1] It is also estimated that this rate increases to 7% by the second year of life.[2]

Children can be born with multiple congenital anomalies that can range from mild to severe to life threatening. Infants born with severe anomalies (e.g., anencephaly, trisomy 13, trisomy 18, or inoperable congenital heart defects) often die. Not all congenital anomalies are serious, and many can be corrected with treatment.

1 Demographics and Risk Indicators Working Group, C.C.A.S.N., Public Health Agency of Canada. Congenital Anomalies Surveillance in Canada: Results of a 2006–2007 Survey on Availability of Selected Data Variables in Canadian Provinces and Territories. Ottawa: Public Health Agency of Canada; 2010.

2 Health Canada. Congenital Anomalies in Canada — A Perinatal Health Report, 2002. Ottawa: Minister of Public Works and Government Services Canada; 2002

5.1.5 Birth Prevalence of Congenital Anomalies, by year, Canada (Excluding Quebec*), 1998 to 2009



*Quebec was excluded because data were not available for all years.

Graphic adapted by CICH from Table B1.1 in 'Congenital Anomalies in Canada 2013: A Perinatal Health Surveillance Report'. Ottawa: Public Health Agency of Canada; 2013.

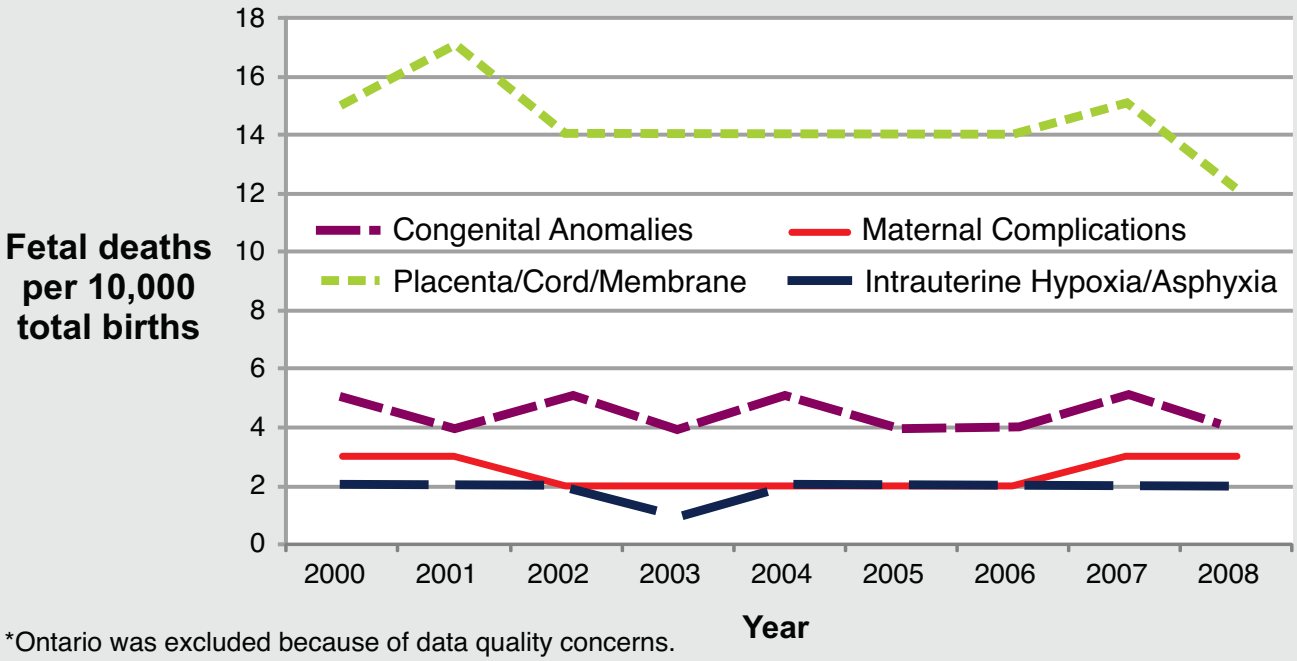
Overall it appears that both the number of babies born with congenital anomalies and the rate per 10,000 total births decreased between 1998 and 2009 in Canada. The rate has decreased from 451.2 per 10,000 total births to 385.2, a decline of approximately 15%. This could be partially due to the decline in some of the common congenital anomalies, such as neural tube defects [view report], but also to inconsistencies in data collection.

It should be noted, however, that some congenital anomalies, such as gastroschisis (where the intestines are outside of the body wall), are increasing in frequency. The precise cause of this is uncertain.[1] The decrease in rates among live births has been noteworthy, leading to birth prevalence below 400 per 10,000 in 2009. On the other hand, the rates of congenital anomalies in stillbirths has increased slightly, primarily because of an increase in the rate among stillbirths of very low birth weight (less than 750 g).[2]

[1] CAPSNet 2012 Annual Report. The Canadian Pediatric Surgery Network. Version 1. February 2013. http://www.capsnetwork.org/portal/Portals/0/CAPSNet/Annual%20Reports/CAPSNet%20AR%202012%20-%20FINAL_Feb%202013.pdf.

[2] Public Health Agency of Canada. Congenital Anomalies in Canada 2013: A Perinatal Health Surveillance Report. Ottawa: Public Health Agency of Canada; 2013

5.2.1 Rate of Fetal Death ≥ 500 Grams by Cause, Canada (Excluding Ontario*), 2000 to 2008

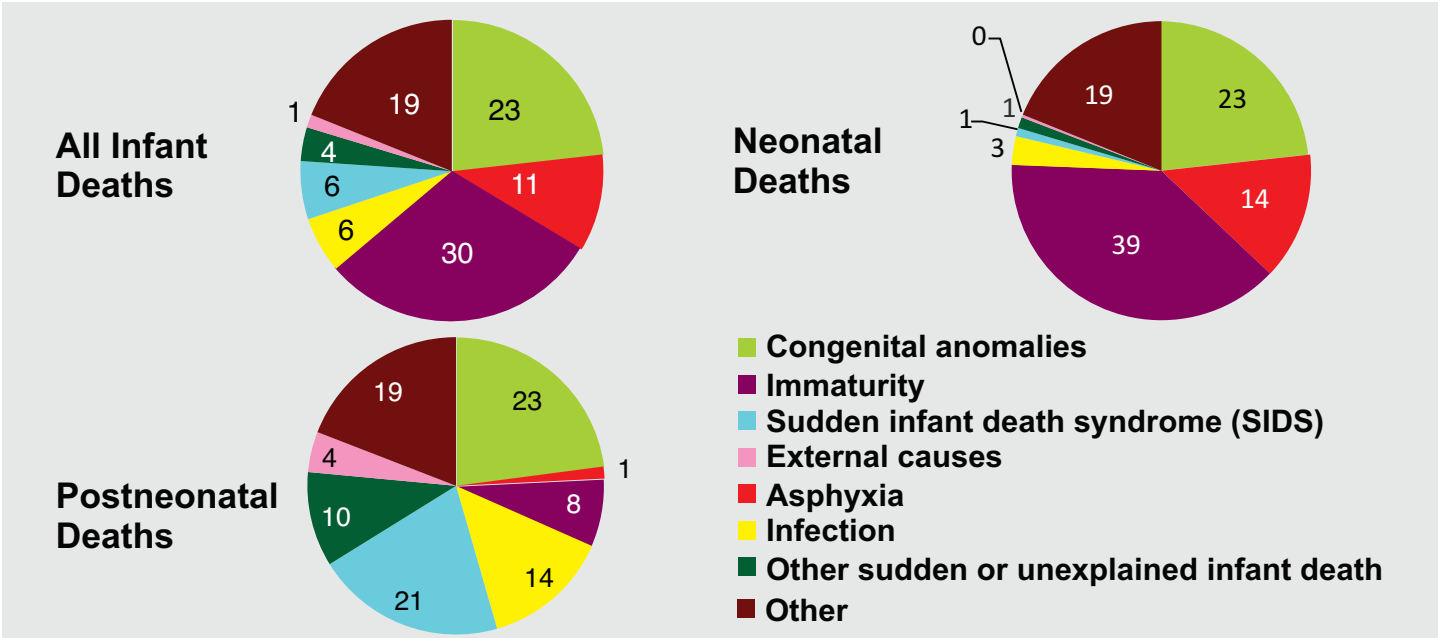


*Ontario was excluded because of data quality concerns.

Graphic created by CICH using data from: Public Health Agency of Canada. Perinatal Health Indicators for Canada 2011. Ottawa, 2012.

Congenital anomalies are the second leading cause of fetal death. Between 2000 and 2008, the rate of fetal death due to congenital anomalies showed little change, varying between 4 and 5 per 10,000 total births.

5.2.2 Causes of Neonatal, Postneonatal, and All Infant Death, Canada (Excluding Ontario*), 2003 to 2007



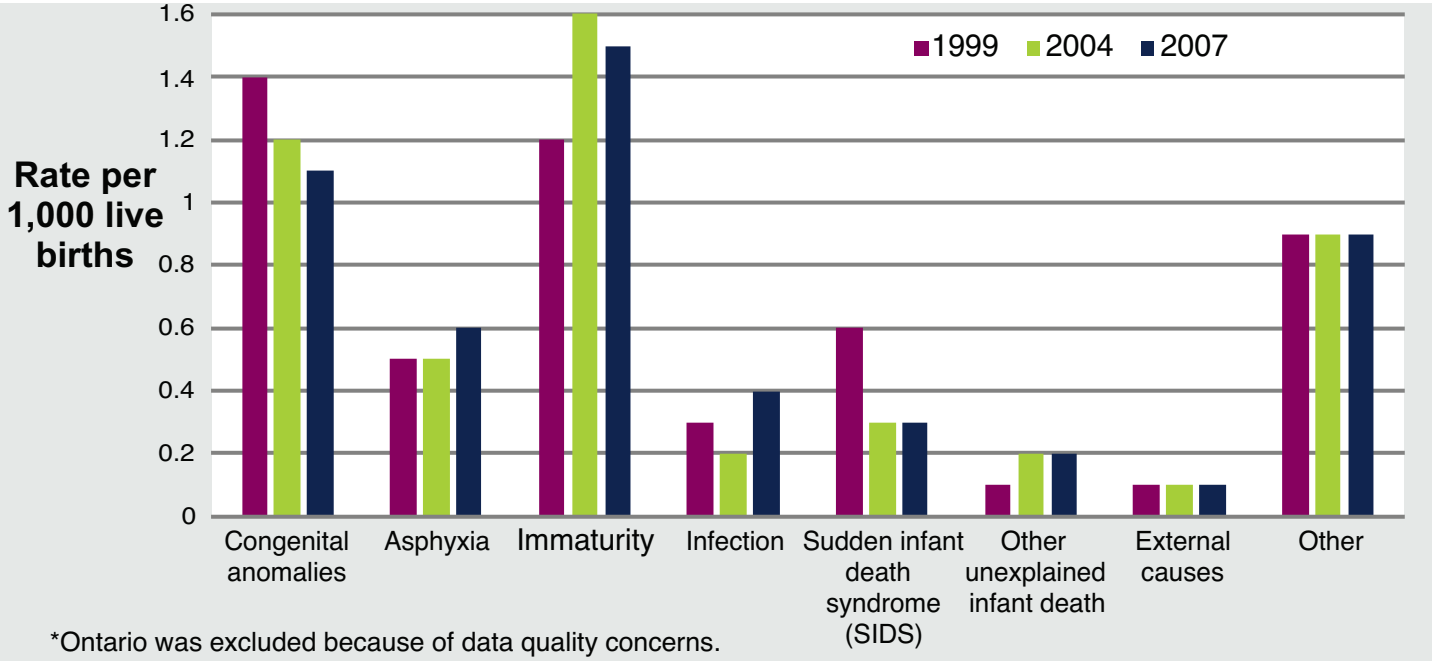
*Ontario was excluded because of data quality concerns.

Graphic created by CICH using data from the Perinatal Health Indicators for Canada 2011. Ottawa: Public Health Agency of Canada; 2012.

Congenital anomalies are a leading cause of infant mortality. They are the leading cause of death in the period from 28 days to one year of age (postneonatal period). They are the second leading cause of death during the first 27 days of life (neonatal period).[1]

[1] Public Health Agency of Canada. Perinatal Health Indicators for Canada 2011. Ottawa: Public Health Agency of Canada; 2012

5.2.3 Causes of Infant Death, Canada (Excluding Ontario*), 1999, 2004, and 2007



*Ontario was excluded because of data quality concerns.

Graphic created by CICH with data from the Public Health Agency of Canada and the Congenital Anomalies Surveillance Network, 2008 (1999 data), and from the Public Health Agency of Canada, Perinatal Health Indicators for Canada 2011. Ottawa, 2012. (data for 2003 to 2007)

Although congenital anomalies are one of the leading causes of infant death in Canada, the rate is decreasing.[1] The Public Health Agency of Canada stated that the decreasing rate of infant deaths is most likely a result of increasing prenatal diagnosis and termination of pregnancies when congenital anomalies are diagnosed.[2]

[1] Public Health Agency of Canada. Perinatal Health Indicators for Canada 2011. Ottawa: Public Health Agency of Canada; 2012.

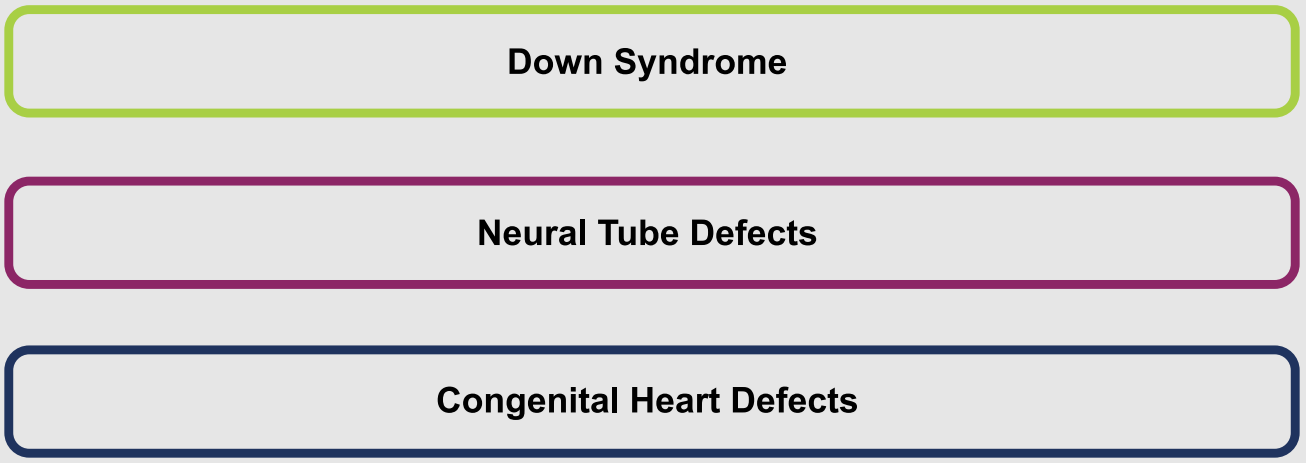
[2] Public Health Agency of Canada and the Congenital Anomalies Surveillance Network, 2008

Implications

It is important to monitor congenital anomalies for possible associations with environmental factors, as the human genome responds to the environment in a very dynamic fashion.[3] In an effort to facilitate health service planning, methods for collecting information regarding congenital anomalies in Canada could be improved for more accessible and reliable data.

[3] Health Canada. Congenital Anomalies in Canada — A Perinatal Health Report, 2002. Ottawa: Minister of Public Works and Government Services Canada; 2002

5.3.1 Three Common Congenital Anomalies



Graphic created by CICH.

There are many congenital anomalies. The module contains some more detailed information on three of the most common congenital anomalies: Down syndrome, neural tube defects, and congenital heart defects.

5.3.2 Common Congenital Anomalies - Down Syndrome

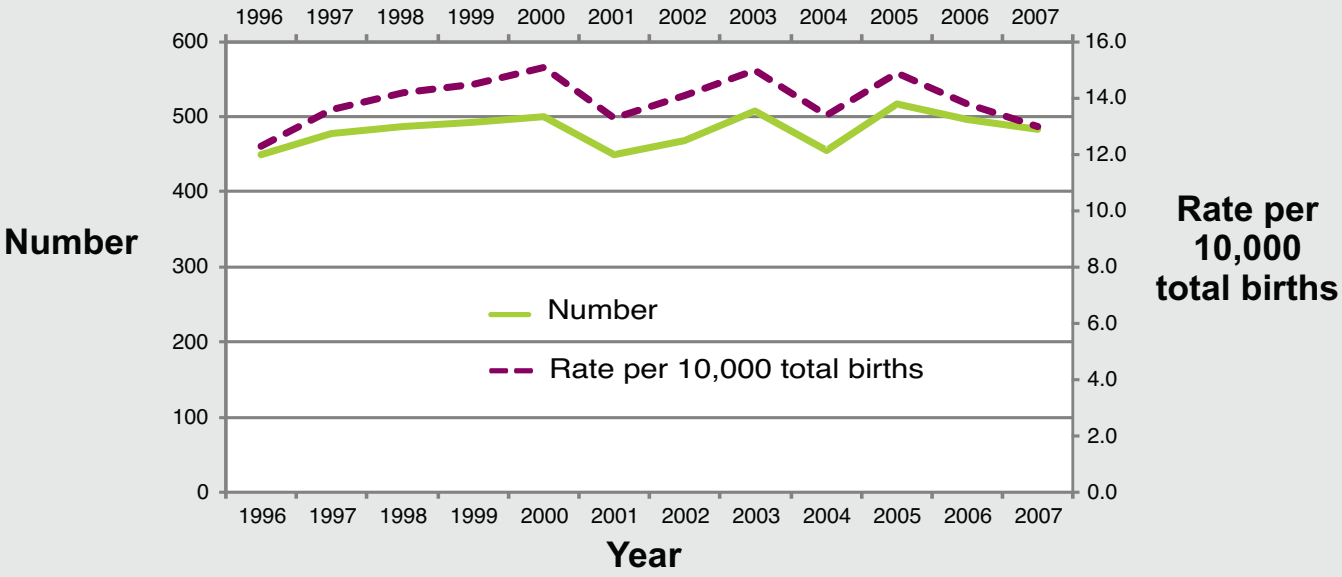
Image credit: Shutterstock

Most children with Down syndrome have an extra copy of the 21st chromosome, meaning that instead of two of these chromosomes, they have three, which is referred to as Trisomy 21. About 95% of people with Down syndrome have Trisomy 21. People with Down syndrome have wide variations in mental abilities, behaviour, and development, and their symptoms can range from mild to severe.[1]

While the chance of having an infant with Down syndrome increases with maternal age, a baby with Down syndrome can be born to women of any age. It is estimated 80% of children with Down syndrome are born to women younger than 35 years of age.[1]

[1] Your Child with Down Syndrome. Canadian Down Syndrome Society. http://www.cdss.ca/images/pdf/brochures/english/your_child_with_down_syndrome_english.pdf

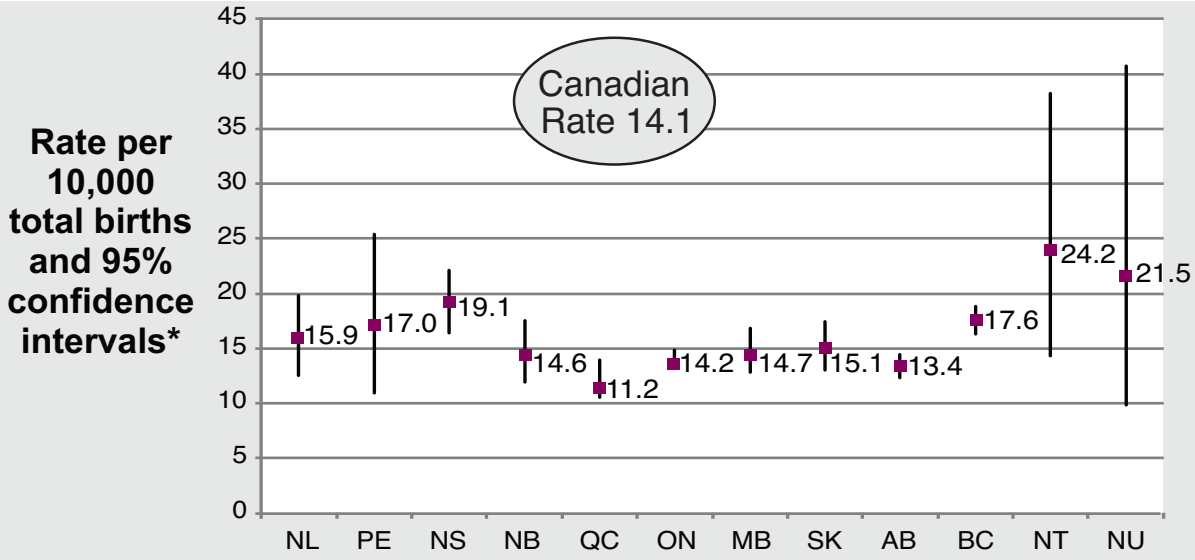
5.3.3 Common Congenital Anomalies - Rates of Down Syndrome, Canada, 1996 to 2007



Graphic created by CICH with data from the 'Perinatal Health Indicators for Canada 2011'. Ottawa: Public Health Agency of Canada; 2012

There has been no clear trend in the rates of Down syndrome since 1996. In 2007, the rate was 13.0 per 10,000 total births, or approximately 483 babies born with Down syndrome.

5.3.4 Common Congenital Anomalies - Rate of Down Syndrome per 10,000 Births, Canada, (Excluding Yukon†), 1998 to 2007



† The rate from Yukon is not shown due to small numbers.

* A confidence interval is "an interval of values bounded by confidence limits within which the true value of a population parameter is stated to lie with a specified probability". (http://dictionary.reference.com/browse/confidence%20interval) In this case, we can be 95% confident that the provincial rates of Down syndrome per 10,000 total births is within the interval shown with the black lines.

Graphic created by CICH using data from 'Perinatal Health Indicators for Canada 2011'. Ottawa: Public Health Agency of Canada; 2012.

When looking at the combined rate of Down syndrome for the years 1998–2007, it is clear that the rate varied greatly between provinces and territories. The rates of Down syndrome ranged from 11.2 per 10,000 total births in Quebec to 24.2 in the Northwest Territories.

These differences may be due to a number of factors, such as differences in maternal age, the availability and use of prenatal screening and diagnosis, and the rate of pregnancy termination.

5.3.5 Common Congenital Anomalies - Neural Tube Defects

Open Neural Tube Defects

Examples:
- **Anencephaly**
- **Encephalocele**
- **Spinal Bifida - the most common type**

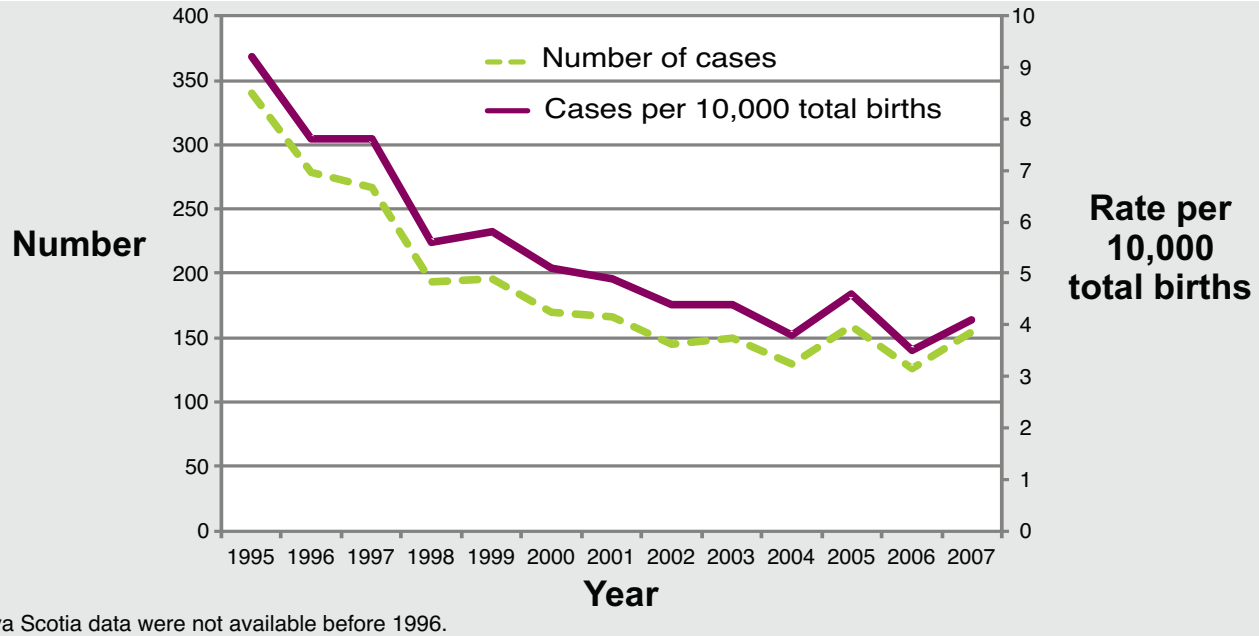
Closed Neural Tube Defects

Examples:
- **Lipomyelomeningocele**
- **Lipomeningocele**
- **Tethered Cord**

Graphic created by CICH using information from http://www.chg.duke.edu/diseases/ntd.html

Neural tube defects are a group of congenital abnormalities of the central nervous system that result when the bony structure that encloses the spinal cord (the vertebra) does not close completely. The causes of neural tube defects are multifactorial and are influenced by geography, ethnicity, genetics, and nutrition.

5.3.6 Common Congenital Anomalies - Rate of Neural Tube Defects, Canada, 1995 to 2007*

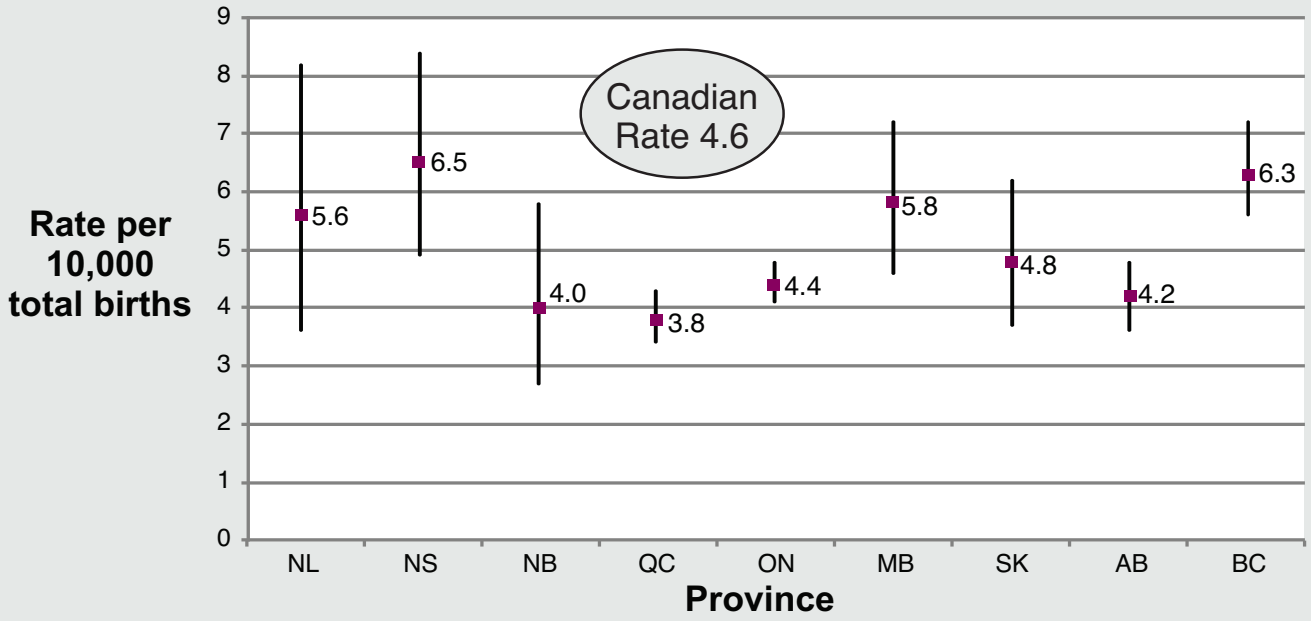


*Nova Scotia data were not available before 1996.

Graphic created by CICH using data from: Perinatal Health Indicators for Canada 2011. Years 1998 to 2007. Ottawa: Public Health Agency of Canada; 2012 and Canadian Perinatal Health Report, 2008 Edition. Years 1995 to 1997. Ottawa: Public Health Agency of Canada; 2008.

In 1999, in Canada, six babies were born with neural tube defects per 10,000 total births, a decline from 11.1 per 10,000 births in 1989. Between 1995 and 2007, the rate of neural tube defects in Canada decreased by about half, to 4.1 from 9.2 per 10,000 total births. Most of the decline occurred between 1995 and 2004, and there has not been a clear trend in rates since. Despite this, much of the decline is attributed to policy-making and education initiatives surrounding the role of folic acid in preventing babies from developing neural tube defects.

5.3.7 Common Congenital Anomalies - Rate of Neural Tube Defects, Canada*, 1998 to 2007



*The rates for Prince Edward Island, Yukon, Northwest Territories, and Nunavut are not released due to small numbers.

Graphic created by CICH using data from: Perinatal Health Indicators for Canada 2011. Ottawa: Public Health Agency of Canada; 2012.

There are variations in the rates of neural tube defects across Canada, ranging from 3.8 per 10,000 total births in Quebec to 6.5 in Nova Scotia.

5.3.8 Common Congenital Anomalies – Neural Tube Defects – Primary Prevention Policies

Image credit: Shutterstock

Research shows that women can reduce their risks of having a baby with a neural tube defect by taking folic acid, or folate, which is a B vitamin. Folic acid is essential for the development of the baby's brain and spine. Studies have shown that women who take enough folic acid supplements and eat a healthy diet before they become pregnant and during the early part of their pregnancy are less likely to have a baby with a neural tube defect.[1]

In 1998, the Canadian government required that white flour, enriched pasta, and cornmeal be fortified with folic acid.[2] The Public Health Agency of Canada and Health Canada recommended that all women who could become pregnant take a supplement containing 0.4mg of folic acid and suggested a higher dose for women who are at increased risk of having a baby with a neural tube defect.[3]

[1] Lumley J, Watson L, Watson M, Bower C. Periconceptional supplementation with folate and/or multivitamins for preventing neural tube defects. Cochrane Database Syst Rev. 2001;3.

[2] Millar W. Folic Acid Supplementation. Statistics Canada, Health Reports (Catalogue 82-003-XIE0). Ottawa: Statistics Canada; 2004;15(3):49–52.

3 Van Allen MI, McCourt C, Lee NS. Preconception health: folic acid for the primary prevention of neural tube defects. A resource document for health professionals, 2002. Ottawa: Minister of Public Works and Government Services Canada; 2002. Catalogue No.: H39-607/2002E

5.3.9 Common Congenital Anomalies - Folic Acid Use Before Pregnancy, Canada, 2009–2010

Women Aged 20 to 49 Who Took Folic Acid Before Their Last Pregnancy, Canada, 2009-2010	Total: 59.8%

Age	
20 to 24 years	33.50%
25 to 29 years	53.20%
30 to 49 years	65.80%

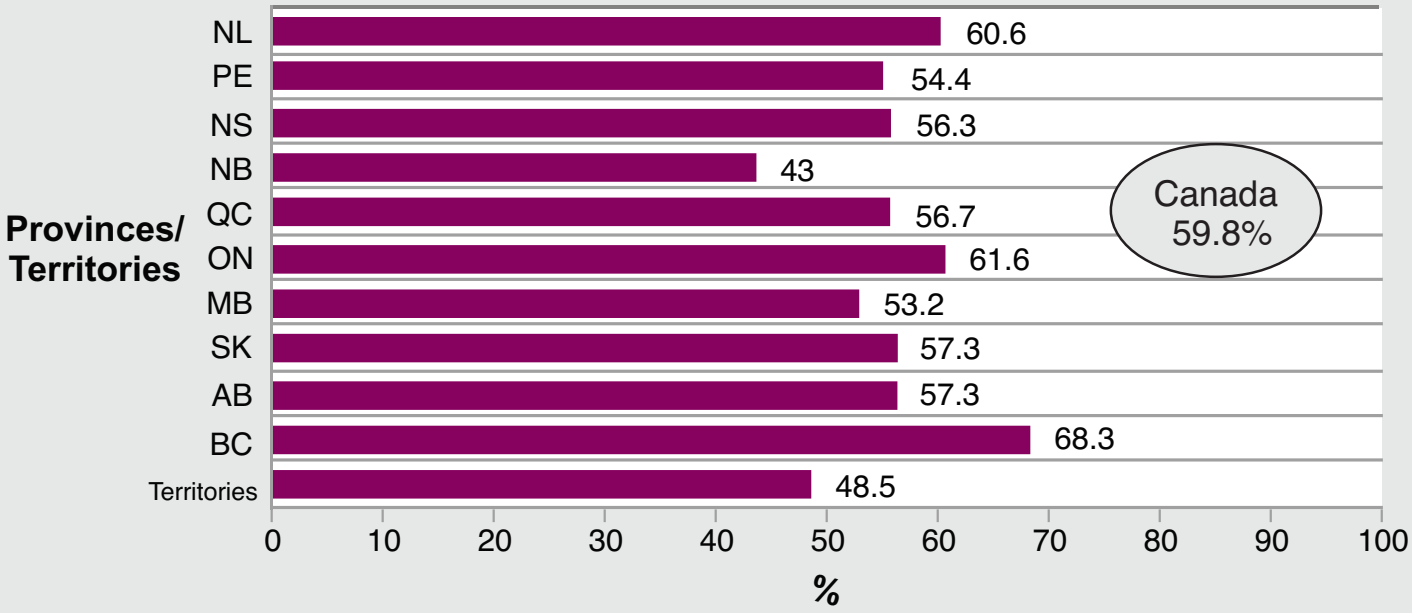
Education	
Less than secondary school graduation	33.70%
Secondary school graduation	44.70%
Some post-secondary	50.00%
Post-secondary graduation	67.20%

Household Income	
Quintile 1	43.30%
Quintile 2	52.00%
Quintile 3	65.10%
Quintile 4	71.70%
Quintile 5	84.20%

Graphic created by CICH using adapted data from the Canadian Community Health Survey PUMF, 2009–2010.

According to the 2009–2010 Canadian Community Health Survey, nearly 60% of women aged 20 to 49 years reported taking folic acid before their last pregnancy. However, access to folic acid and prevention of neural tube defects is not equal for all women. Women aged 30 to 49 years were more likely to take folic acid than were younger women. Two-thirds of women with a post-secondary education took the supplements compared to one-third of those without a high school diploma. Moreover, 84% of women in the highest income households took folic acid compared with only 43% of women in the poorest households.

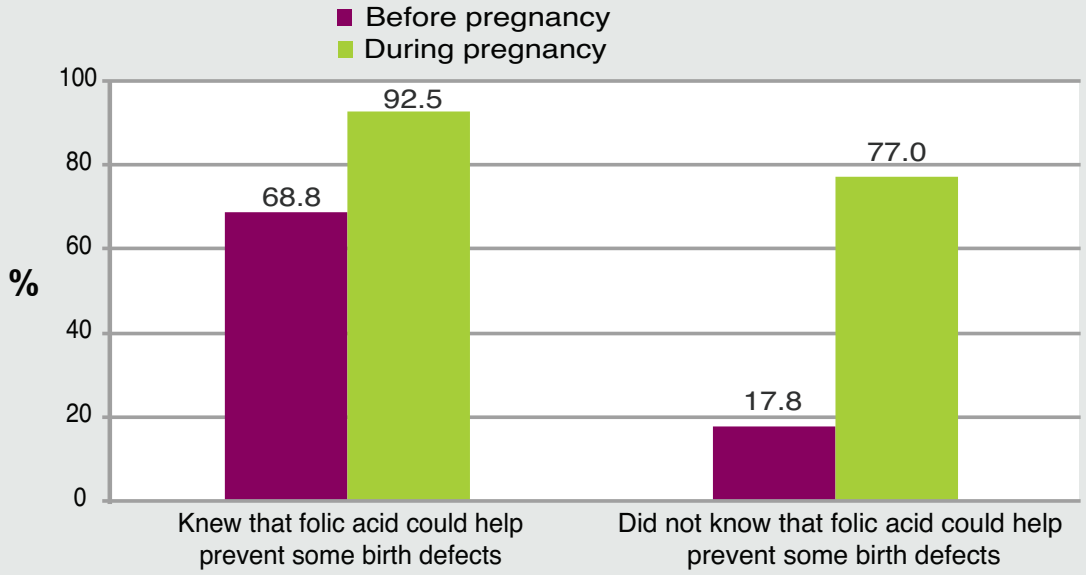
5.3.10 Common Congenital Anomalies – Women Aged 20 to 49 Years Who Took Folic Acid Before their Last Pregnancy, Canada, 2009–2010



Graphic created by CICH using adapted data from the Canadian Community Health Survey PUMF, 2009–2010.

There are also differences among the provinces and territories with regards to the proportion of women taking folic acid prior to pregnancy. Approximately 49% did so in the territories compared with 68% in British Columbia.

5.3.11 Common Congenital Anomalies – Women Who Took Folic Acid by Pre-Pregnancy Knowledge, Canada*, 2006–2007



* 6,421 birth mothers 15 years of age and older who had a single live born baby during the three-month period preceding the 2006 Canadian Census of Population and lived with their baby. The rates for Prince Edward Island, Yukon, Northwest Territories, and Nunavut are not released due to small numbers.

Graphic created by CICH using data from: What Mothers Say: The Canadian Maternity Experiences Survey. Ottawa: Public Health Agency of Canada; 2009.

According to the Canadian Maternity Experiences Survey, more than three-quarters of women in the survey knew before their pregnancy that folic acid could help prevent some birth defects. Women who knew that this was the case were more likely to take folic acid before and during their pregnancy.[1]

[1] Public Health Agency of Canada. What Mothers Say: The Canadian Maternity Experiences Survey. Ottawa: Public Health Agency of Canada; 2009

5.3.12 Common Congenital Anomalies – Congenital Heart Defects

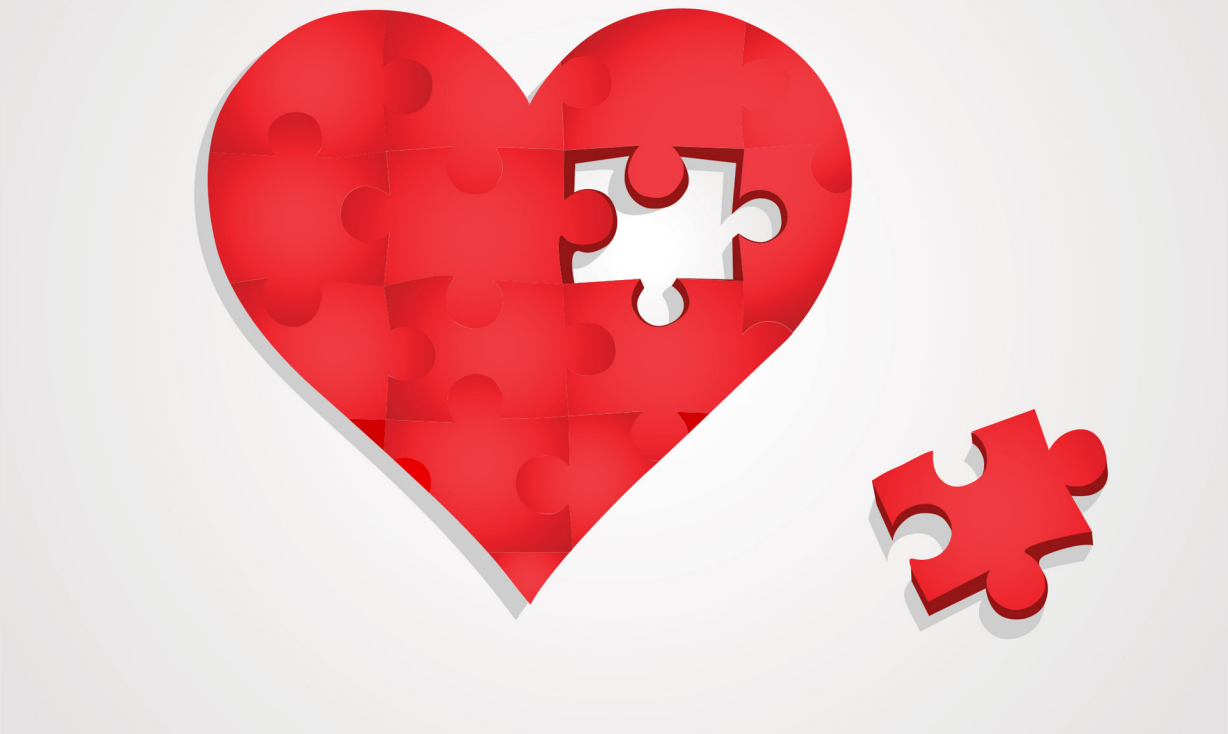


Image credit: Shutterstock

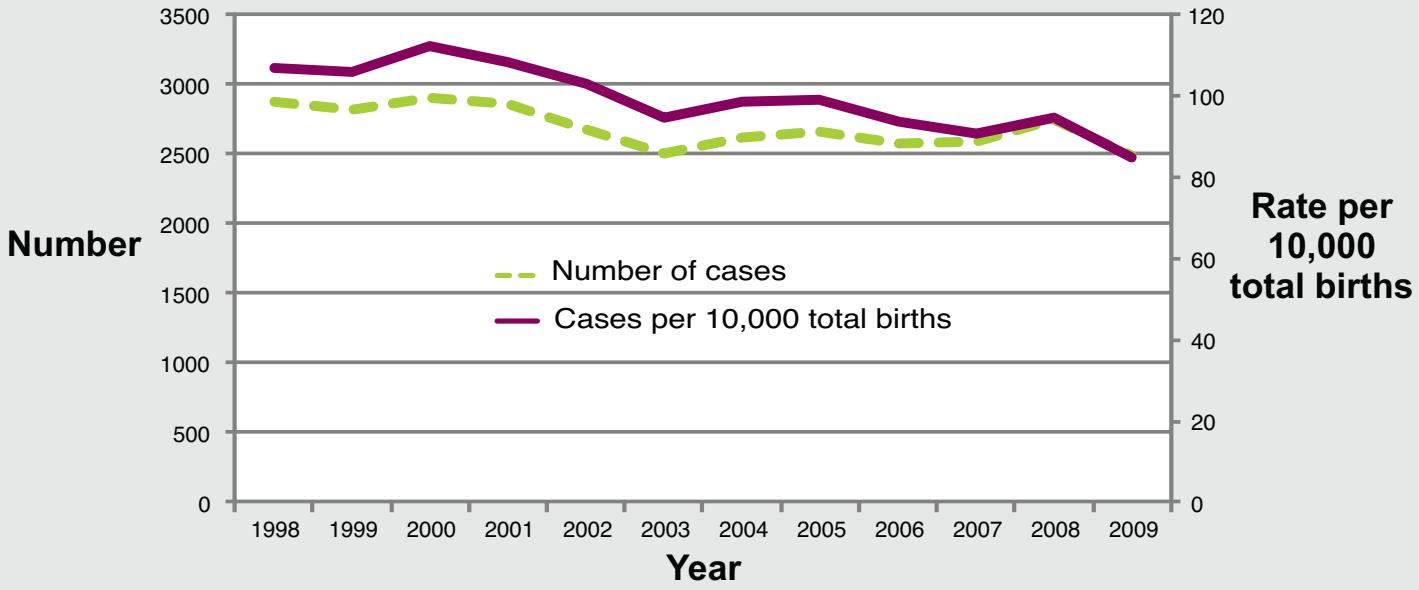
Congenital heart defects – where the heart or the blood vessels near the heart do not develop normally before birth – are the most common congenital anomalies. In Canada, 1 in 100 to 150 babies are born with a congenital heart defect.[1] There has been progress in the early diagnosis and surgical treatment of congenital heart defects that has resulted in a decrease in death and illness. Sixty years ago, less than 20% of infants born with complex heart defects lived to adulthood. Today, more than 90% live to adulthood due largely to surgical procedures, the development of regional cardiac surgical centres, and improved medical care.[2] Despite this progress, congenital heart defects remain the leading cause of childhood death among congenital anomalies in Canada. Children who suffer from the most serious heart defects require complex medical care and can have a greatly compromised quality of life.[1]

In most situations, the cause of congenital heart defects is unknown. Although many congenital heart defects can be genetic, viral infections such as rubella (measles) or drug and/or alcohol use during pregnancy are known to increase risks. Thus, the causes are multifactorial. Babies born with congenital anomalies often have other congenital or chromosomal anomalies, such as Down syndrome, trisomy 13 or 18, or Turner syndrome.[2]

[1] Health Canada. Congenital Anomalies in Canada — A Perinatal Health Report, 2002. Ottawa: Minister of Public Works and Government Services Canada; 2002.

[2] Congenital Heart Defects. Heart and Stroke Foundation of Canada http://www.heartandstroke.com/site/c.ikIQLcMWJtE/b.3484063/

5.3.13 Common Congenital Anomalies – Rate of Congenital Heart Defects, Canada (Excluding Quebec*), 1998–2009

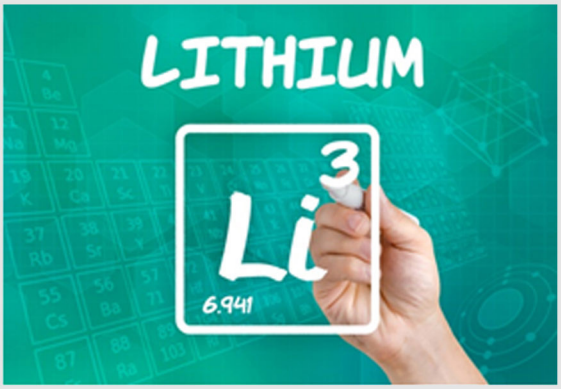


*Quebec was excluded because data were not available for all years.

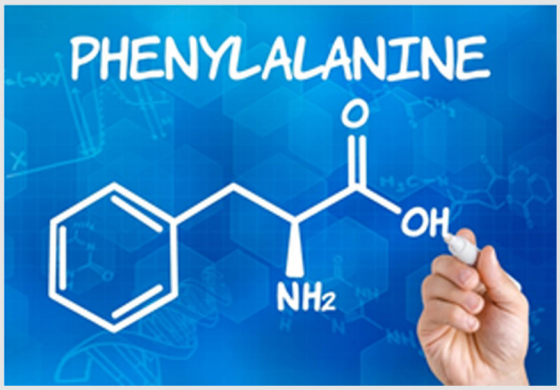
Graphic created by CICH using data from Table B4.1 in 'Congenital Anomalies in Canada 2013: A Perinatal Health Surveillance Report'. Ottawa: Public Health Agency of Canada; 2013.

Both the number and the rate of congenital heart defects (determined up to 30 days of age) have decreased. In 1998, the rate was 107.1 per 10,000 total births. By 2009, that rate had decreased by 21% to 85.1 per 10,000 total births.

6.1.1 Introduction to Teratogens



What do recreational drugs, lithium, phenylalanine, and chicken pox have in common?



They are all examples of possible teratogens.

Image credit: Shutterstock

The human epigenome responds to the environment in a very dynamic fashion. For example, toxic exposure, stress, diet, and other factors influence chemical "switches" that control gene expression. Teratogens are one of these factors.

A teratogen is a substance known to cause birth defects following exposure during pregnancy. Some teratogens can be drugs (e.g., prescription drugs such as lithium or epilepsy medication or recreational drugs). Certain infections, such as rubella (German Measles) or chicken pox can also be teratogens. The mother can also unknowingly introduce the fetus to teratogens in the womb. For example, in the case of PKU, a mother with PKU herself, who does not follow the prescribed diet, has very high levels of phenylalanine that passes to her fetus through placental circulation and leads to congenital anomalies in the fetus, such as congenital heart disease.

It is often geneticists and genetic counsellors who provide counselling for mothers and infants exposed to teratogens.

6.1.2 Alcohol

Image credit: Best Start: http://www.beststart.org/resources/alc_reduction/pdf/static_clings_bi_lr.pdf

Alcohol is a common teratogen. Alcohol use during pregnancy can adversely affect the unborn baby. There are many factors that influence this effect, including the amount of alcohol ingested over time and differences in the way the mother metabolizes alcohol. There is also evidence that variations in a person's genetic makeup can affect the baby's susceptibility to alcohol while in utero.[1]

The effects of alcohol exposure during pregnancy are considered on a spectrum, and thus are called fetal alcohol spectrum disorder (FASD). These effects can include physical, mental, behavioural, and learning disabilities, as well as cognitive, emotional, and behavioural issues.[2]

The medical diagnoses of FASD include Fetal Alcohol Syndrome (FAS), Partial FASD (pFAS), and Alcohol-Related Neurodevelopmental Disorder (ARND).[3] The diagnosis is always related to restriction in growth, changes in facial features, problems with the central nervous system, brain damage, and prenatal exposure to alcohol.[4]

[1] Reynolds JN, Weingbert J, Clarren S, Beaulieu C, Rasmussen C, Kobor M, et al. Fetal alcohol spectrum disorders: gene-environment interactions, predictive biomarkers, and the relationship between structural alterations in the brain and functional outcomes. Semin Pediatr Neurol. 2011;18(10):49–55

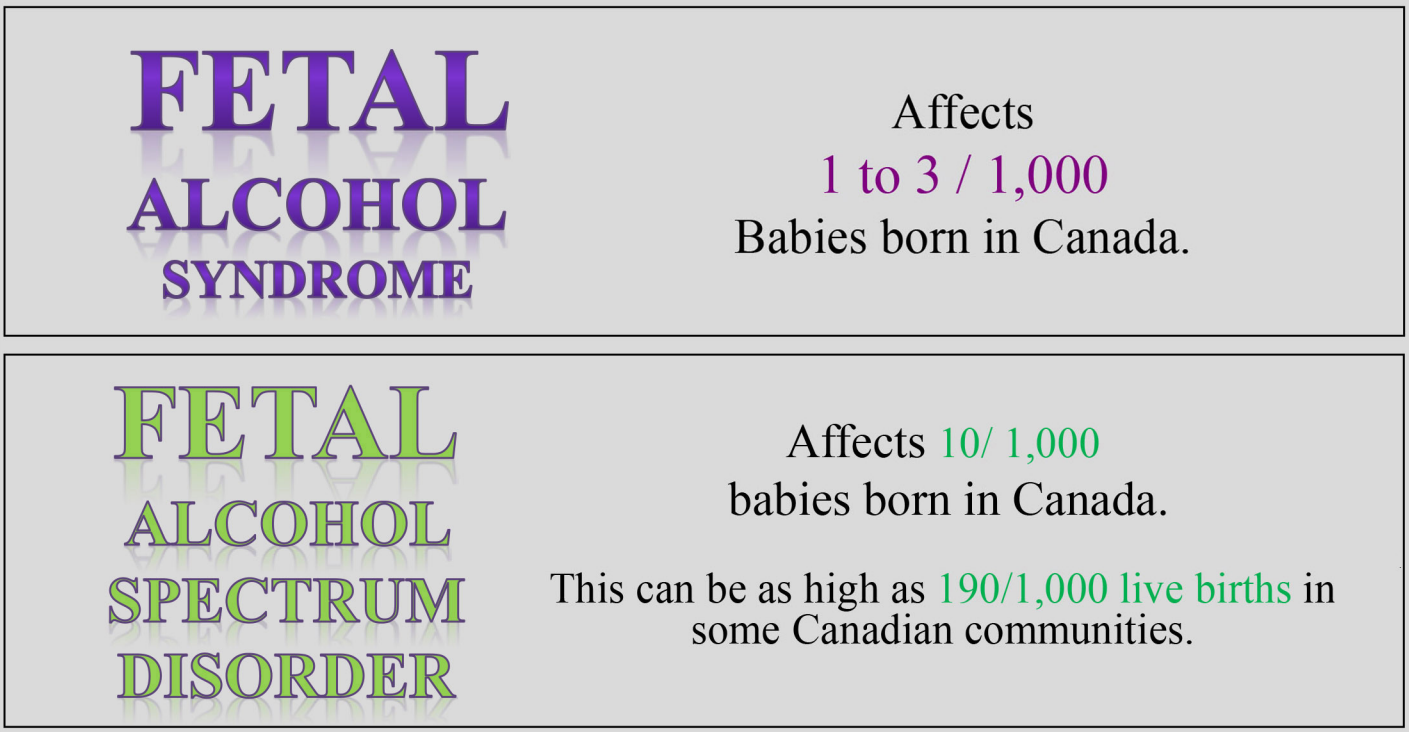
[2] Chudley AE, Conry J, Cook JL, Loock C, Rosales T, LeBlanc N. Fetal alcohol spectrum disorder: Canadian guidelines for diagnosis. CMAJ. 2005;172(5 suppl)

[3] Public Health Agency of Canada, Fetal Alcohol Spectrum Disorder (FASD). http://www.phac-aspc.gc.ca/hp-ps/dca-dea/prog-ini/fasd-etcaf/index-eng.php

[4] Carson G, Cox LV, Crane J, Croeau P, Graves L, Kluka S, et al. Alcohol Use and Pregnancy Consensus Clinical Guidelines. SOGC Clinical Practice Guideline. JOGC. 2010; 32(8). http://sogc.org/wp-content/uploads/2013/01/gui245CPG1008E.pdf

MOTHERISK, at the Hospital for Sick Children, is an excellent resource for more information on alcohol and pregnancy, and the effects of other drugs and substances during pregnancy. MOTHERISK offers The Alcohol and Substance Use Helpline (1-877-327-4636), which provides information and counselling to pregnant and breastfeeding women, their families, and healthcare providers.

6.1.3 Fetal Alcohol Spectrum Disorder – The Facts



Graphic created by CICH using date from Robinson GC, Conry JL, Conry RF. Clinical profile and prevalence of fetal alcohol syndrome in an isolated community in British Columbia. CMAJ 1987;137:203–7.

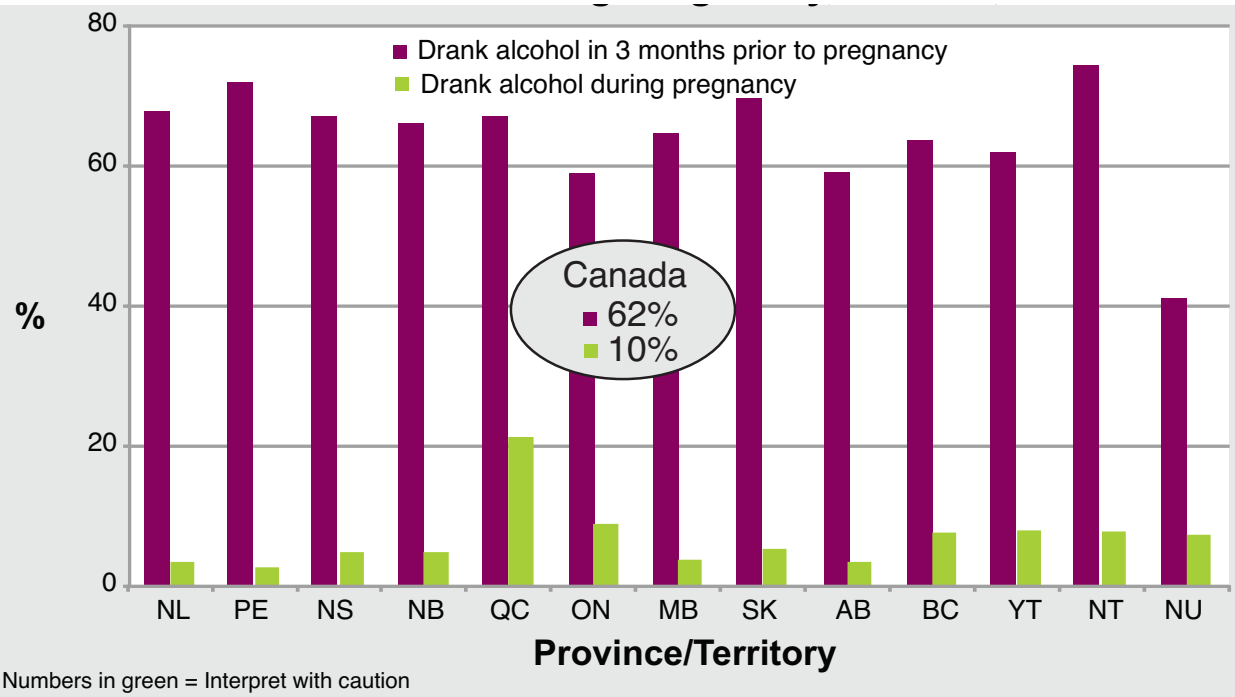
It is estimated that 1 to 3 per 1,000 babies born in Canada have fetal alcohol syndrome (FAS). Fetal alcohol spectrum disorder (FASD) is estimated to affect approximately 1% of the population. Some communities in Canada report prevalence rates as high as 190 per 1,000 live births.[1]

[1] Robinson GC, Conry JL, Conry RF. Clinical profile and prevalence of fetal alcohol syndrome in an isolated community in British Columbia. CMAJ. 1987;137:203–7

Implications

FASD cannot be cured. It can have lifelong effects on individuals, their families, and their communities. There is a need to develop a better understanding of risk factors associated with FASD.

6.1.4 Fetal Alcohol Spectrum Disorder – Alcohol Use During Pregnancy, Canada, 2009



Numbers in green = Interpret with caution

Graphic created by CICH using data from the Public Health Agency of Canada. What Mothers Say: The Canadian Maternity Experiences Survey. Ottawa, 2009.

According to a national survey of new mothers, 62.4% of women reported drinking alcohol during the three months before their pregnancy. However, only 10.5% of women reported consuming alcohol during pregnancy, and that number varied by province and territory. Less than one percent (0.7%) of mothers reported drinking frequently. Furthermore, 11% indicated they had engaged in binge drinking before realizing they were pregnant.[1]

[1] Public Health Agency of Canada. What Mothers Say: The Canadian Maternity Experiences Survey. Ottawa: Public Health Agency of Canada; 2009

6.1.5 Fetal Alcohol Spectrum Disorder – Primary Prevention

FASD is the leading cause of preventable developmental disability among Canadians. Because health behaviours must be considered within the greater context of the lives of women and their families, women require a variety of support, education, and policy approaches to enable them to maintain their health. A number of national groups in Canada have endorsed evidence-based practice guidelines – Alcohol Use and Pregnancy Consensus Clinical Guidelines – to help practitioners support women around alcohol use.[1]

[1] Endorsed by: The Society of Obstetricians and Gynaecologists of Canada; Motherisk; The College of Family Physicians of Canada; Canadian Association of Midwives; Association of Obstetricians and Gynecologists of Quebec; Federation of Medical Women of Canada; Society of Rural Physicians of Canada; Canadian Association of Perinatal and Women's Health Nurses.

Alcohol Use and Pregnancy: Consensus Clinical Guidelines

- Universal screening for alcohol consumption should be done periodically for all pregnant women and women of child-bearing age.
- Healthcare providers should create a safe environment for women to report alcohol consumption.
- The public should be informed that alcohol screening and support for women at risk is part of routine women's healthcare.
- Healthcare providers should be aware of the risk factors associated with alcohol use in women of reproductive age.
- Brief interventions are effective and should be provided by healthcare providers for women with at-risk drinking.
- If a woman continues to use alcohol during pregnancy, harm reduction/treatment strategies should be encouraged.
- Pregnant women should be given priority access to withdrawal management and treatment.
- Healthcare providers should advise women that low-level consumption of alcohol in early pregnancy is not an indication for termination of pregnancy.

Source: Carson G, Cox LV, Crane J, Croeau P, Graves L, Kluka S, et al. Alcohol Use and Pregnancy Consensus Clinical Guidelines. SOGC Clinical Practice Guideline. JOGC. 2010;32(8). http://sogc.org/wp-content/uploads/2013/01/gui245CPG1008E.pdf

6.1.6 Fetal Alcohol Spectrum Disorder – Primary Prevention

Graphic created by CICH using a graphic from Best Start, http://www.beststart.org/resources/alc_reduction/index.html

It is essential that women and families have accurate information about drinking alcohol during pregnancy.

Some Myths and Facts about Alcohol and Substance Use

MYTH: Alcohol or drugs taken after the first trimester will not affect the unborn baby.

FACT: Most organ development is completed a few weeks after the first trimester. Brain development continues throughout pregnancy and after birth. Exposure to substances any time in the pregnancy can affect the baby's brain.

MYTH: One drink in pregnancy is enough to harm the unborn baby.

FACT: A safe amount of alcohol in pregnancy is not known. It is unlikely, though, that a single drink before a mother knew she was pregnant could damage her unborn baby.

MYTH: There is no hope for a baby exposed to heavy drug and alcohol use.

FACT: There is always hope. Drug and alcohol use in pregnancy affects each baby differently.

Source: MOTHERISK, http://www.motherisk.org/women/alcohol.jsp#two

7.1.1 Genetic Testing and Screening – The Growth in Genetic Testing

There are currently genetic tests for more than 3,500 conditions.

New discoveries are being made at a very rapid rate.

Image credit: Shutterstock. Data from the National Institutes of Health. Genetic Testing Registry. http://www.ncbi.nlm.nih.gov/gtr/ 2012.

The demand for new genetic tests and genetic services is growing exponentially, largely because they are more readily available and affordable than ever before. In addition, genetic testing can now be used for both rare and common conditions.[1] Requests for genetic tests and related services are increasing for chronic conditions, such as cardiovascular diseases, developmental delay, dysmorphic features, neurological disorders (e.g., neonatal seizures), and eye diseases (e.g., retinoblastoma).

In 2012, the Canadian Institutes of Health Research and Genome Canada announced a $65 million program to support research projects in the fields of genomics and personalized medicine. This could result in the development of new genetic tests for Canadians.[2]

Which health professionals are ordering genetic tests is also changing. Once exclusively the job of medical geneticists, now family doctors, oncologists, neurologists, cardiologists, haematologists, ophthalmologists, microbiologists, and pathologists frequently order genetic tests.[1]

[1] McMaster Health Forum. Evidence Brief: Coordinating the Use of Genetic Tests and Related Services in British Columbia. Hamilton, Canada: McMaster Health Forum; 2012.

[2] Canadian Institutes of Health Research. Harper government invests in personalized medicine. Canadian Institutes of Health Research; 2012. http://www.cihr-irsc.gc.ca/e/44825.htm

Implications

These trends in expanding genetic testing capabilities call for greater coordination and streamlined service delivery to determine which tests should be provided, where, and by whom. The increased availability of, and demand for, genetic tests and related services will put increased pressure on current programs and services, especially for common diseases and conditions.[1]

7.1.2 Genetic Screening – Nuchal Translucency Screening in Pregnancy

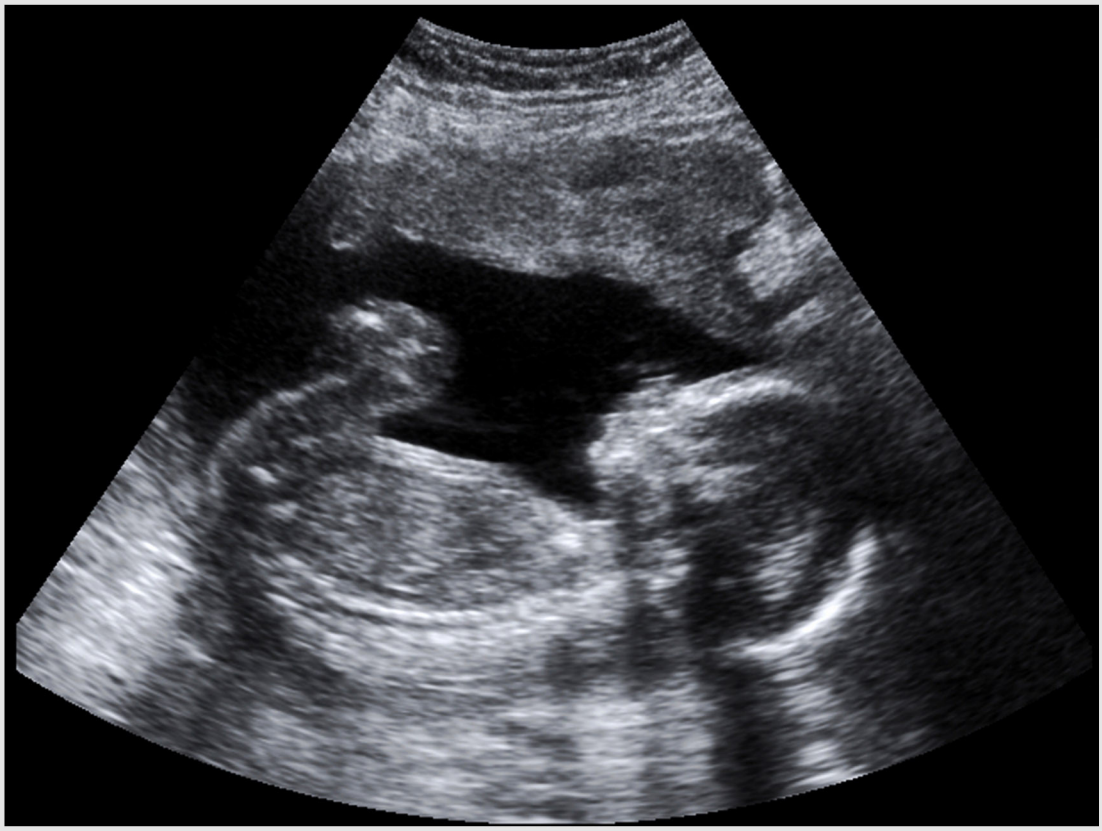


Image credit: Shutterstock

Nuchal translucency screening uses ultrasound to screen for Down syndrome, other conditions caused by an extra chromosome (trisomy 13 and 18), and congenital heart defects. It is performed between 11 and 14 weeks of pregnancy. When nuchal translucency screening is done with blood tests, the results are more accurate. Because nuchal translucency is a screening procedure, it provides a risk estimate for developing one of the conditions. A follow-up diagnostic test – such as chorionic villus sampling or amniocentesis – confirms the diagnosis.

The Society of Obstetricians and Gynaecologists of Canada and the Canadian College of Medical Geneticists recommend that nuchal translucency should only be done where the ultrasound staff is specially trained and accredited to do this procedure. Therefore it is not available in all communities across Canada.[1]

[1] Joint SOGC-CCMG Clinical Practice Guideline. No. 261 (Replaces No. 187, February 2007). Prenatal Screening for Fetal Aneuploidy in Singleton Pregnancies. J Obstet Gynaecol Can. 2011;33(7):736–50. http://www.sogc.org/guidelines/documents/gui261CPG1107E.pdf

7.1.3 Consent and Newborn Genetic Screening

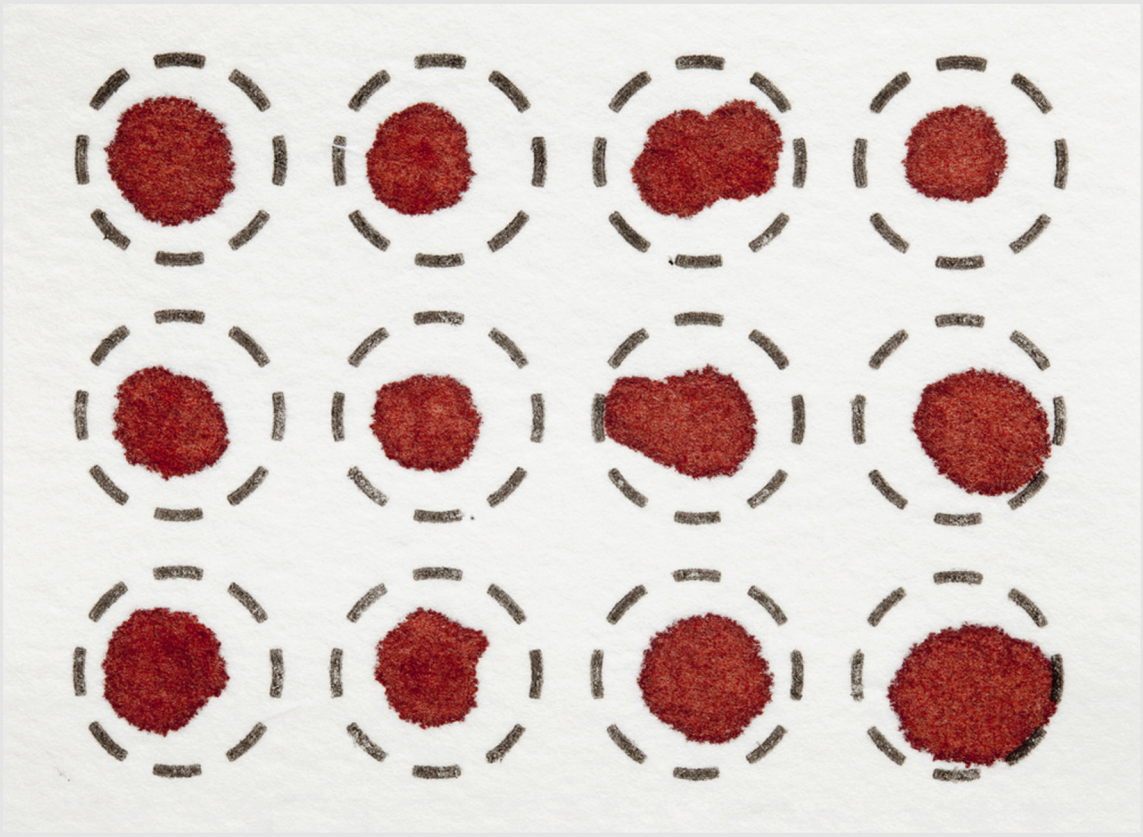


Image credit: Shutterstock

Anecdotal and published evidence about newborn screening programs show that many parents are not aware that babies are offered a screening test. This issue has sparked controversy and debate about the issue of obtaining consent.

Most newborn screening programs do not require explicit consent from parents. Newborn screening is considered part of routine healthcare for children. Newborn screening for treatable and preventable conditions is considered to be in the best interests of the child. In Canada, we have what is called "implied consent," which means parents are not necessarily asked. Doctors assume the parents want screening unless they say otherwise. As a result, although parents are provided with newborn screening pamphlets and the right to decline testing, they may not be aware that newborn screening has been carried out and for which diseases.

Implications

Newborn screening raises a number of legal and ethical issues. Parents may have limited knowledge of the programs and, for this reason, the informed consent process provides an opportunity to educate them on the specifics of the screening procedures. Requiring explicit consent, however, requires additional resources for program implementation. Nevertheless, the timing for giving this information is important.

7.1.4 New Technologies and Newborn Genetic Screening

Image credit: Shutterstock

New technologies are now available that allow screening for a large number of conditions through a single process. One technology that is moving into the clinic to help with diagnosis and treatment is whole genome sequencing (WGS).[1,2] Of note, however, the level and use of new technologies varies from site to site and between jurisdictions. It is not readily available everywhere, therefore it is not accessible to all children and families.

[1] Green RC, Berg JS, Grody WW, Kalia SS, Korf BR, Martin CL, et al. ACMG Recommendations for Reporting of Incidental Findings in Clinical Exome and Genome Sequencing. American College of Medical Genetics and Genomics; 2013. http://www.acmg.net.

[2] Saunders CJ, Miller NA, Soden SE, Dinwiddie DL, Noll A, Alnadi NA, et al. Rapid whole-genome sequencing for genetic disease diagnosis in neonatal intensive care units. Sci Transl Med. 2012;4(154):154ra135

7.1.5 Newborn Screening: Important Questions

Image credit: Shutterstock

As the technological frontier expands, it is possible that whole genome sequencing may supplant newborn screening programs. Before doing so, there are many important questions:

- How will we make decisions about the use of this technology and how will we evaluate its use?
- Who decides which conditions will be screened for?
- What criteria and evidence will be used to decide which conditions to screen for?
- How will these programs be implemented?

7.1.6 Genetic Testing and Screening: Whole Genome Sequencing (WGS)

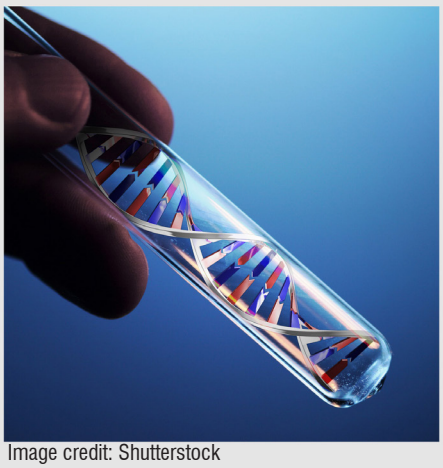


Image credit: Shutterstock

A person's genome is his/her complete set of DNA. Each human genome contains all of the information needed to build and maintain that individual. A copy of the entire genome – which has more than 3 billion DNA base pairs – is contained in all human cells that have a nucleus.

Whole genome sequencing (WGS) compares large amounts of genetic data to identify variations in DNA associated with specific diseases. Once new genetic associations are identified, researchers can use the information to develop better strategies to detect, treat, and prevent disease.[1]

For a more thorough description of whole genome sequencing, click here.

[1] National Genome Research Institute. National Institutes of Health. http://www.genome.gov/20019523

Implications

The use of WGS in the clinic raises a number of questions. Analyzing the entire genome at once can reveal what are called "incidental" findings. Incidental findings are pieces of information learned when conducting genetic testing that do not relate to the clinical problem or concern at hand. Also, there are many genetic mutations that are not adequately understood and cannot be interpreted. This raises debates about WGS: its usefulness in the clinical setting, communicating results to families, informed consent stipulations, and the right to know or not know.

- WGS challenges current policies with regard to the genetic testing of children. Traditional guidance for genetic testing of children recommends that results revealing conditions of adult onset should not be communicated unless disclosure could prevent serious harm to the health of the child's parents or family members.[2]
- WGS raises questions about potential harms, stigmatization, and discrimination. For example, the potential use of this information by insurance companies.
- The rapidly evolving volume of new information is combined presently with a lack of expertise to interpret and communicate this information. It is important that children not be caught up in the current uncertainty surrounding WGS and the communication of genetic research results and incidental findings.

[2] Arbour L. Guidelines for genetic testing of healthy children. A joint statement with the Canadian College of Medical Geneticists Bioethics Committee, Canadian Paediatric Society (CPS) Ethics and Public Policy Committee, Canadian College of Medical Geneticists. Paediatrics & Child Health. 2003;8(1):42–5. Reference No. B03-01. Reaffirmed January 2011. Addendum (April 2008). http://www.cps.ca/english/statements/B/b03-01.htm

7.1.7 Genetic Testing and Screening – Whole Genome Sequencing (WGS) – Questions for Clinicians

Image credit: Shutterstock

Dealing with such a huge amount of data provides a new set of questions for clinicians:

- How do you evaluate the clinical usefulness and the benefit to patients? Should physicians or clinical laboratories provide genomic information that has no medical importance but is of social or personal consequence to the child (e.g., genes associated with athletic or musical ability)?
- Does whole genome sequencing require a different level of consent than other genetic tests or medical assessments?
- Should patients be informed of results that do not have direct implications for them but do for other family members?
- Should other family members be informed of findings that have direct implications for them that were found on analysis of a relative's genome sequence?
- Do physicians or clinical laboratories have a duty to re-contact patients if sequence data that were previously obtained are later found to have serious medical implications?

7.2.1 Pharmacogenetics

Image credit: Shutterstock

People may react in different ways to drugs based on their genetic makeup. Pharmacogenetics is the study of the how genetic factors influence a person's response to drugs. Testing for certain genetic tendencies before prescribing certain drugs could help avoid harmful drug effects and improve the effectiveness of drug therapy. Pharmacogenetic testing is beginning to revolutionize prescription practices in medicine. The case of codeine is but one example.

Codeine and Breastfeeding: Example of Pharmacogenetic Impact

Some women may produce much more morphine when taking codeine than most people do. In this situation, newborns might be exposed to toxic levels of morphine when breastfeeding.

Health Canada advises the public, especially nursing mothers, about the very rare but serious health risk to breastfed babies posed by codeine use.[1,2] Considering many women receive codeine for post-labour pain, they and their physicians need to be aware of the pharmacogenetic impact of codeine. Options to reduce this risk include discontinuing codeine after 2 to 3 days of use and being aware of symptoms of producing more morphine in both mothers and newborns.

[1] Use of Codeine Products by Nursing Mothers Advisory 2008-164. October 8, 2008. http://healthycanadians.gc.ca/recall-alert-rappel-avis/hc-sc/2008/13255a-eng.php

[2] Madadi P, Moretti M, Djokanovic N, Bozzo P, Nulman I, Ito S, et al. Guidelines for maternal codeine use during breastfeeding. Can Fam Physician. 2009;55(11):1077–8

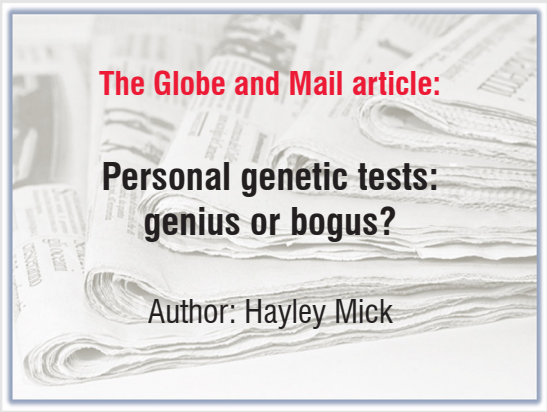
Implications

It will take time for pharmacogenetic testing to enter conventional medical care. Future pharmacogenetic research in children is essential. Children are not small adults. They have unique physiologic differences not only based on their genotype but also on their developmental stage and other factors such as BMI (body mass index). All of these factors will have an effect on their responses to drugs. Therefore adult research is not necessarily transferable to children.

This understanding of pharmacogenetics not only maximizes therapeutic effects but also increases the likelihood that pharmacogenetic differences in children may further complicate clinical trial development for orphan diseases and be a disincentive for drug development targeted to children. As a result, children would not benefit from the promise of pharmacogenetic developments.[3] American and European legislation has put provisions in place in the hope of promoting the development of treatment for children and orphan disease groups. Health professionals, including doctors, nurses, genetic counsellors, and pharmacists, have limited knowledge about pharmacogenetics and will need to receive training to be prepared with the necessary skills for introducing pharmacogenetic testing into their clinical practice.

[3] Joly Y, Sillon G, Silverstein T, Krajinovic M, Avard D. Pharmacogenomics: Don't Forget the Children. Current Pharmacogenomics and Personalized Medicine. 2008;6:77–84

7.2.2 Genetic Tests Sold Directly to Consumers



Graphic created by CICH using a Shutterstock image.

To view this article by Hayley Mick, please click **here**.

Private commercial companies are advertising and selling kits via the internet that allow people to send biological samples for DNA analysis. This is called "direct-to-consumer genetic testing." Consumers receive information about their likelihood of developing certain condition(s) linked to particular genes. Many of these companies conduct genetic testing in children and youth,[1] and the tests are often offered without medical supervision.

Companies use promotional statements such as "let your DNA help you plan for the important things in life" or "our goal is to empower you with genetic insights to help motivate you to improve your health."[2]

While some genetic tests are well validated, some have not been validated or are considered inappropriate for the public.

In Canada, Cepmed (Centre for Excellence in Personalized Medicine) and DNA Direct have developed a Personalized Medicine Portal that provides tools to help patients understand how genetic testing can be used in making treatment decisions and promotes communication between patients and healthcare providers. The Portal provides information about access to specific genetic tests in each province.[3]

1 Howard HC, Avard D, Borry P. Are the kids really all right? Direct-to-consumer genetic testing in children: Are company policies for testing minors clashing with professional norms? European Society of Human Genetics. 2011;19(11):1122–6

2 Kolor K, et al. Health care provider and consumer awareness, perceptions, and use of direct-to-consumer personal genomic tests, United States, 2008. Genetics in Medicine. 2009;11:85–95

See, for example, Navigenics' Terms and Conditions: "you should not interpret Your Report or any other Content as recommending any specific treatment plan, product or course of action. You should always consult your physician or other qualified health provider before starting any new treatment."

http://www.navigenics.com/visitor/what_we_offer/our_policies/terms_conditions/ Accessed August 17, 2011

3 Cepmed launches online personalized medicine portal. Marketwire. February 22, 2012. http://finance.yahoo.com/news/cepmed-launches-online-personalized-medicine-204000631.html

Implications

Direct-to-consumer genetic testing for children may provide useful genetic information, though without the benefit of appropriate genetic and clinical counselling. The Canadian College of Medical Genetics and the Canadian Pediatric Society have established professional norms concerning genetic testing of children. They state that for genetic conditions that will not present until adulthood (susceptibility or predictive testing), testing should be deferred until the child is competent to decide whether they want the information. Many argue that testing children using direct-to-consumer services deprives them of the right to decide for themselves whether they want to receive this genetic information. Therefore, it is not in keeping with these norms.

8.1.1 Summary

Image credit: Shutterstock

The science of genetics and its clinical applications are multidimensional as a result of the mix between genetics, the environment, social, and epigenetic factors. We are beginning to understand the role of some of the factors – for example, in childhood cancers – but the future delivery of genetic services will require much more research and better integration of genetic knowledge into healthcare.

Inherited genetic conditions are critically important in the clinical care of children. The World Health Organization has stated that up to 40% of hospital care of children may be related to children with monogenic conditions (those conditions which result from a modification to a single gene).[1] Another study reports that more than 50% of paediatric hospitalizations were children with genetic conditions or children with conditions with underlying genetic components.[2] The lack of Canadian data about the disease burden of paediatric genetics poses a major challenge considering the growing demand for services, the variation in services provided, and competition for healthcare resources. To this end, it is crucial to collect and evaluate relevant data to assess the impact of genes in combination with behavior, environment, and diet on child health.

According to the 1989 Convention on the Rights of the Child, which guides our policy and healthcare approaches in Canada, the best interests of the child are paramount. To conform to the best interests of the child, certain considerations are in order when using genetic tests. Some address the relevance of treatment and prevention and others focus on the confidentiality of children's genetic test results. If a genetic test reveals the presence of a harmful mutation and there are treatments or effective preventive measures that can be initiated during childhood, it could be clearly beneficial to the actual or future health of the child. If there is no treatment or effective preventive measures that could benefit the actual or future health of the child, there should be no disclosure of the test results (either to the child or to the parents) because it would not be in the best interests of the child. Disclosure would contravene the child's rights of confidentiality and privacy and could have psychosocial consequences.

In order to give children the "best start" possible, prenatal and newborn screening programs are needed across Canada. The goal of newborn screening is to promote infant and child health by identifying babies with treatable conditions as early as possible in order to prevent death, disability, or serious health problems. The lack of national standards for newborn screening in Canada means that access to screening is not equitable for all families across the country.

Given the rapid pace of scientific advances in the field of genetics combined with the use of whole genome sequencing tests in clinical settings, there is a need to build awareness of new genomic technologies among physicians, nurses, and other healthcare providers, as each plays a unique role in the genetics healthcare team. In addition, families require access to accurate information in order to make informed decisions. They will require timely access to clinical geneticists, genetic counsellors, help lines, and support services. Such services are essential to ensure that families considering genetic testing receive up to date information about the tests, including the benefits, their accuracy, the associated risks, and the conditions that can and cannot be identified.

The evolution of the field of genetics provides us with opportunities to enable Canadian children to reach their optimum status of health and wellbeing. It also raises a number of challenges that must be addressed – in a thoughtful and evidence-based manner – to ensure the health and rights of our children are protected.

1 World Health Organization. Genes and Human Disease. Monogenic Diseases.
http://www.who.int/genomics/public/geneticdiseases/en/index2.html

2 McCandless SE, Brunger JW, Cassidy SB. The Burden of Genetic Disease on Inpatient Care in a Children's Hospital. J Hum Genet. 2004;74(1):121–7