Genetics and Paediatric Health: Section 4

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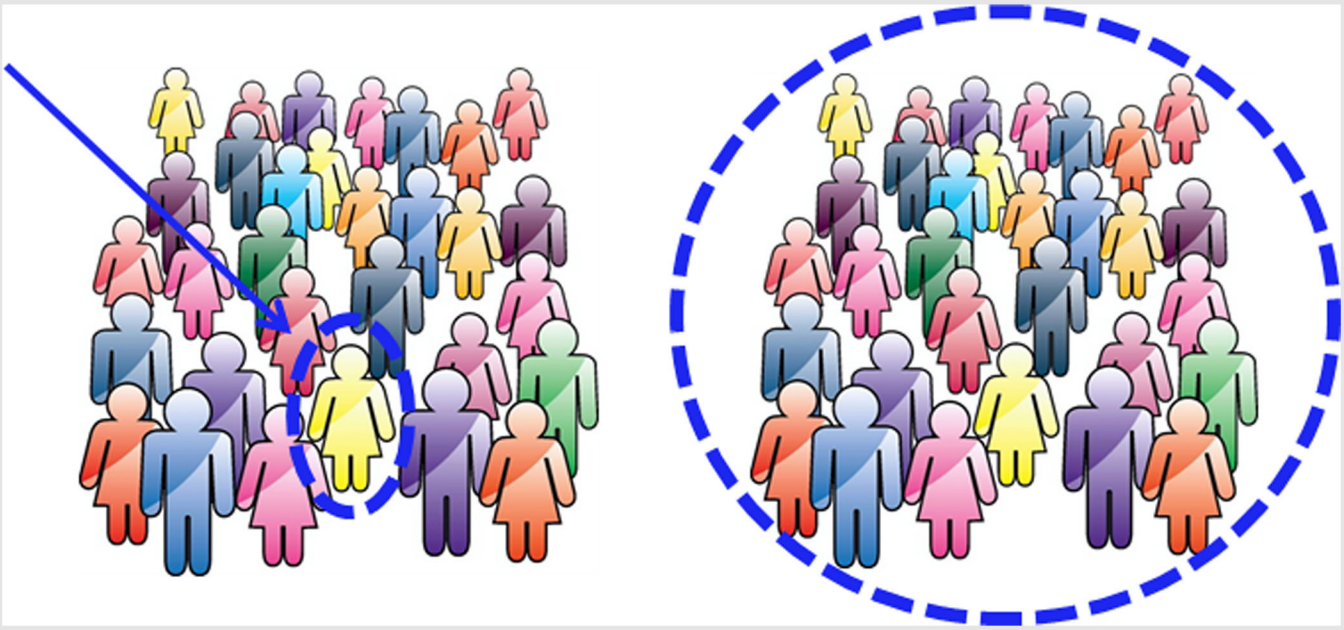
À l'écoute de notre santé

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.

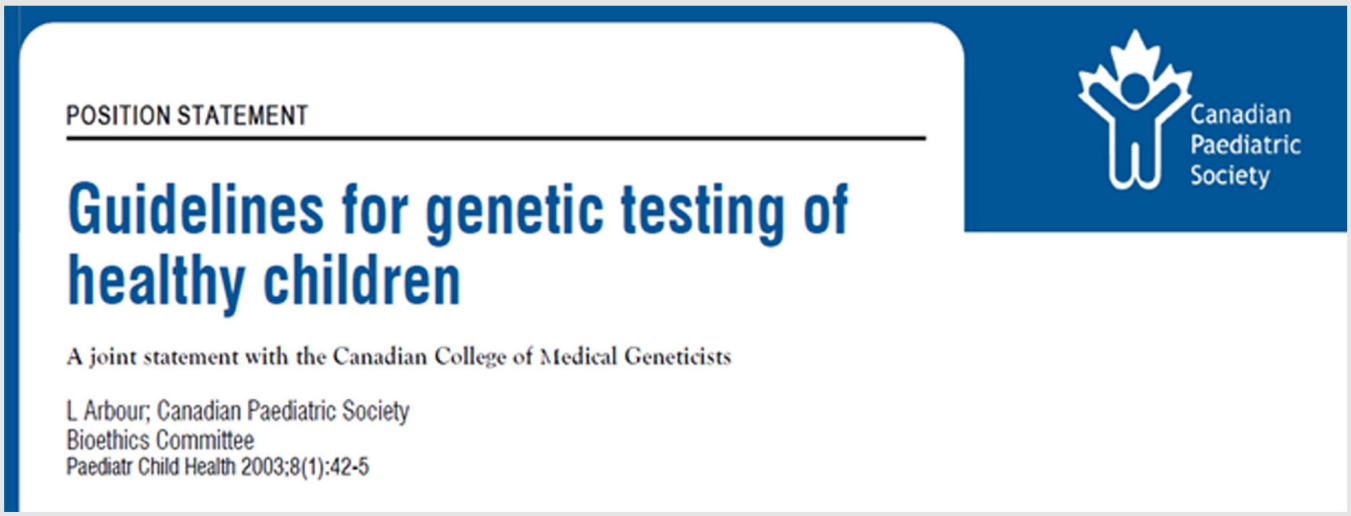
4.2.1 Genetic Testing in Children

Image credit: Shutterstock

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.

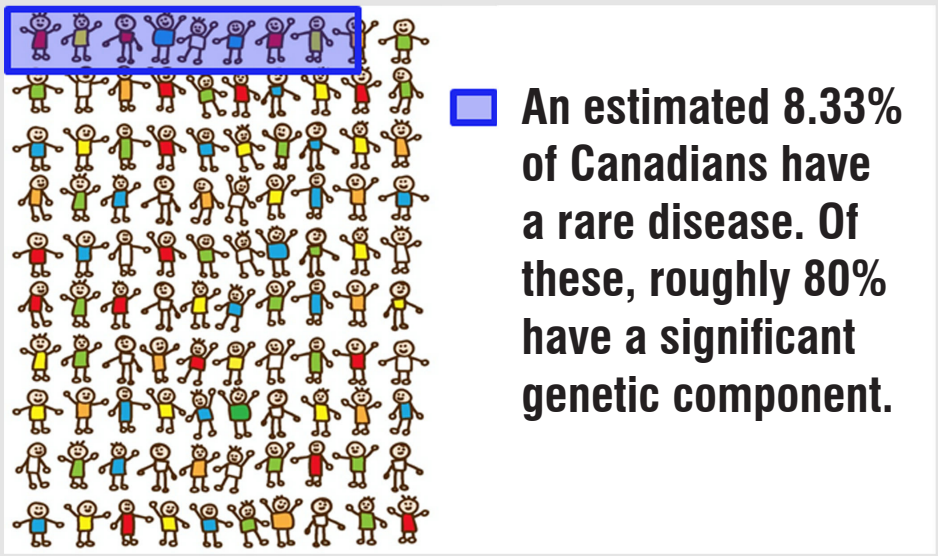
Highlights of the Guidelines for Genetic Testing of Healthy Children[1]

- 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



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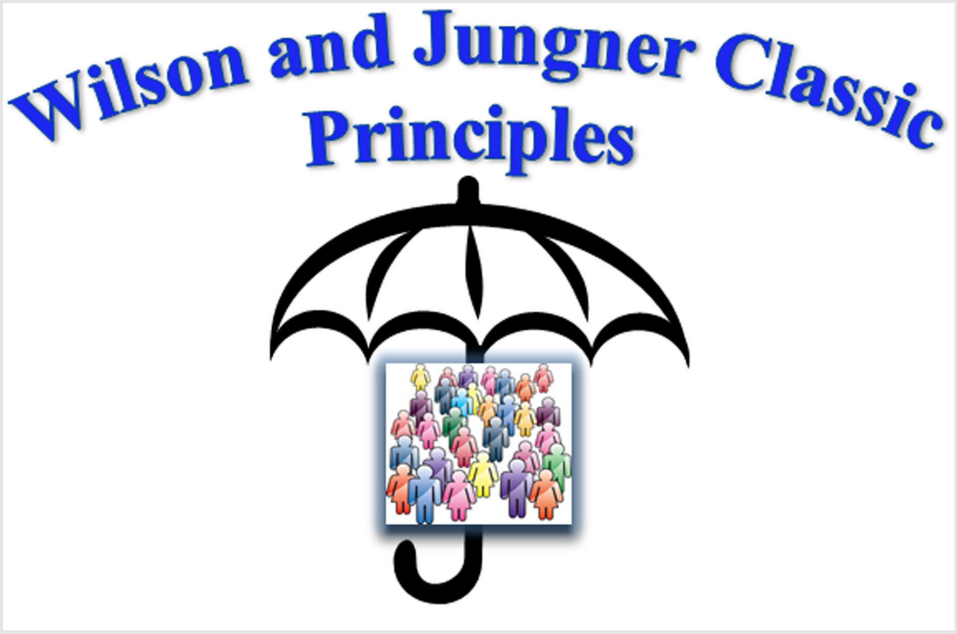
Screening may provide an early signal of a health problem in a child or adult who is not sick at the time of testing per se. It does not provide a definitive diagnosis, but rather suggests that he or she should receive further testing. There are different approaches to genetic screening depending on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

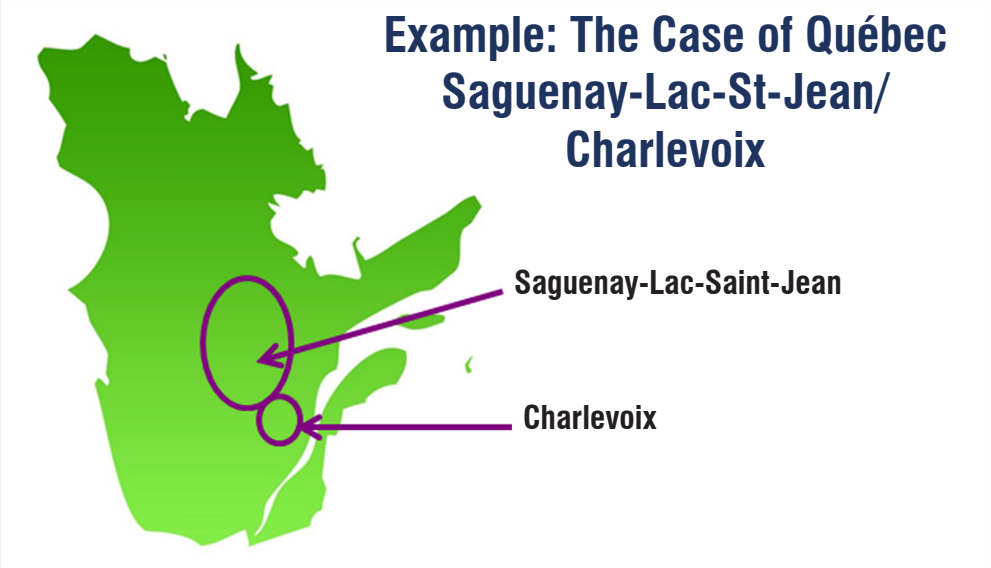
- Family history of an inherited disorder (e.g., a familial chromosome rearrangement).
- Diagnosis during in vitro fertilization prior to implantation.
- Diagnosis prior to artificial insemination.
- People from a specific ethnic group (e.g., thalassemia in people of Mediterranean and Southeast Asian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

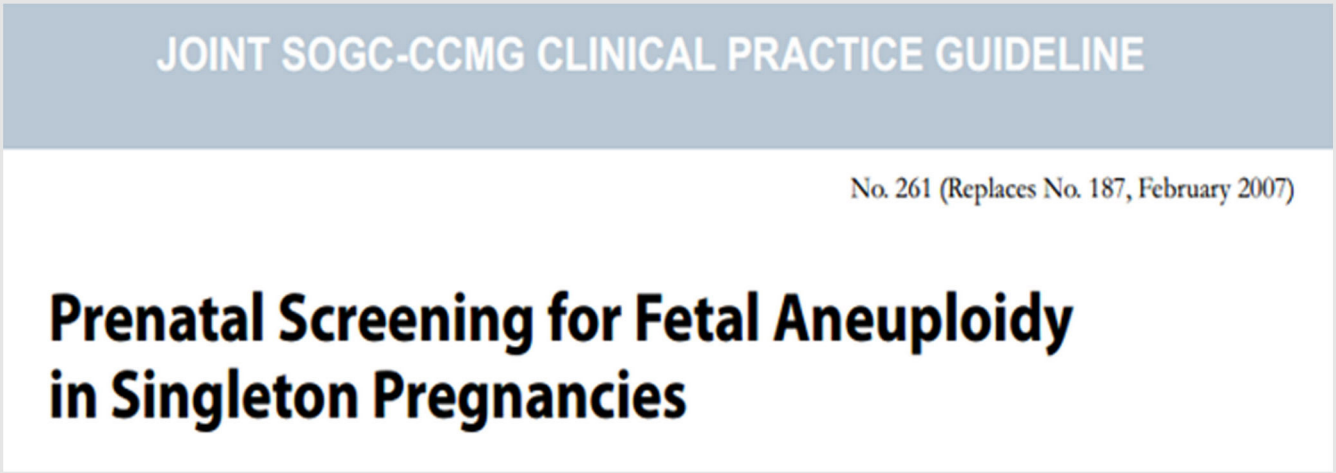
- A higher than expected number of people with ancestry from this region are carriers of one of four autosomal recessive genetic conditions: Autosomal recessive spastic ataxia of Charlevoix-Saguenay; Leigh syndrome, French-Canadian type; Tyrosinemia Type I; and Agenesis of the Corpus Callosum with Peripheral 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.

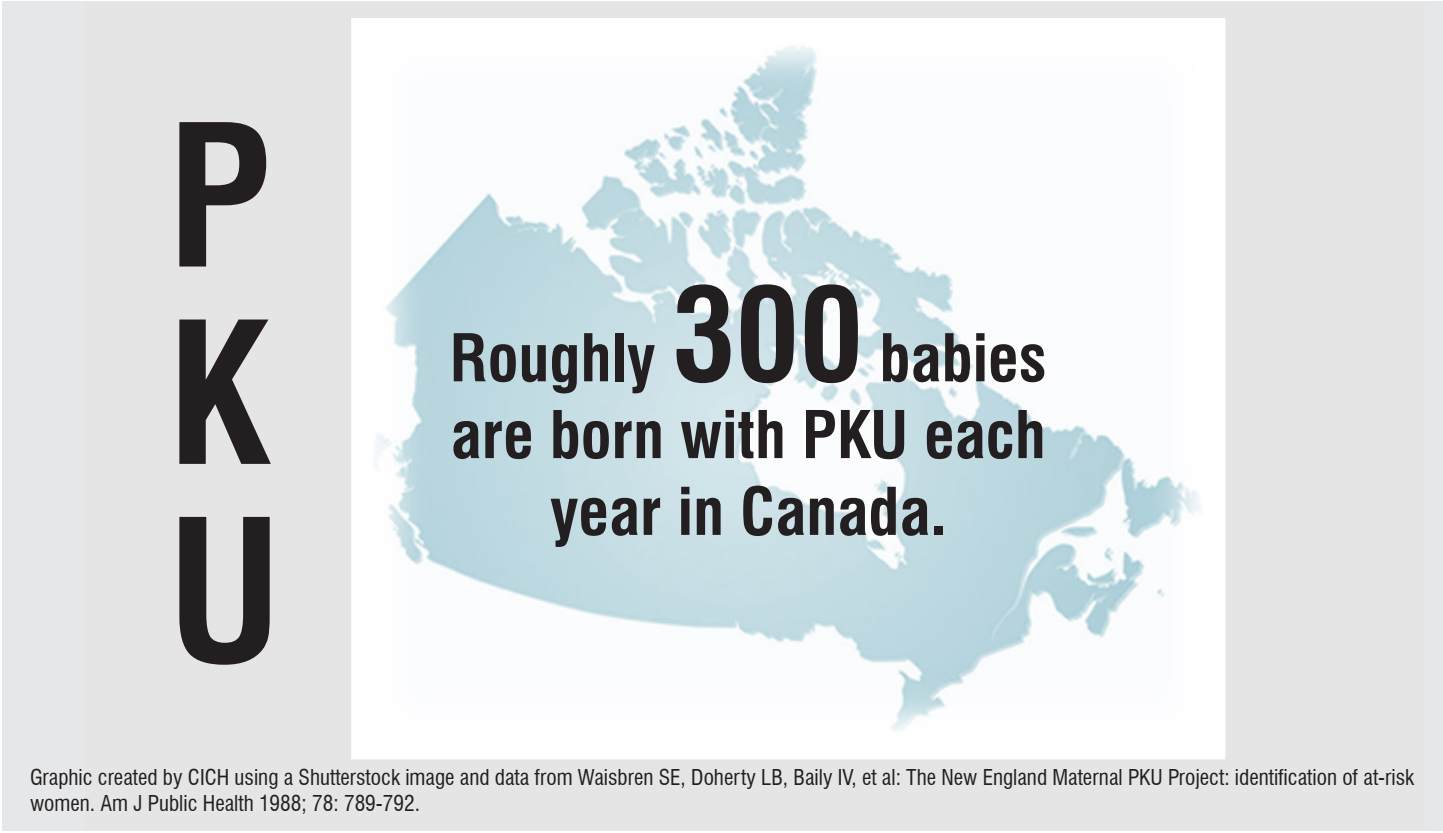
Consent for newborn screening is usually "implied," but parents have the right to opt out of newborn screening. Most provinces provide guidelines for formal documentation when the parents opt out of newborn screening.

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

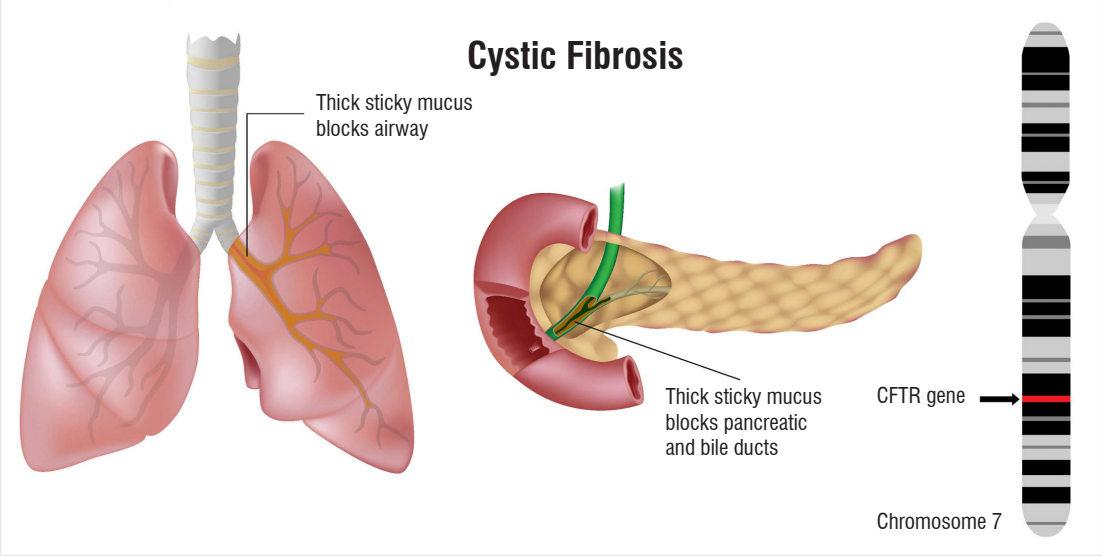


Image credit: Shutterstock

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]

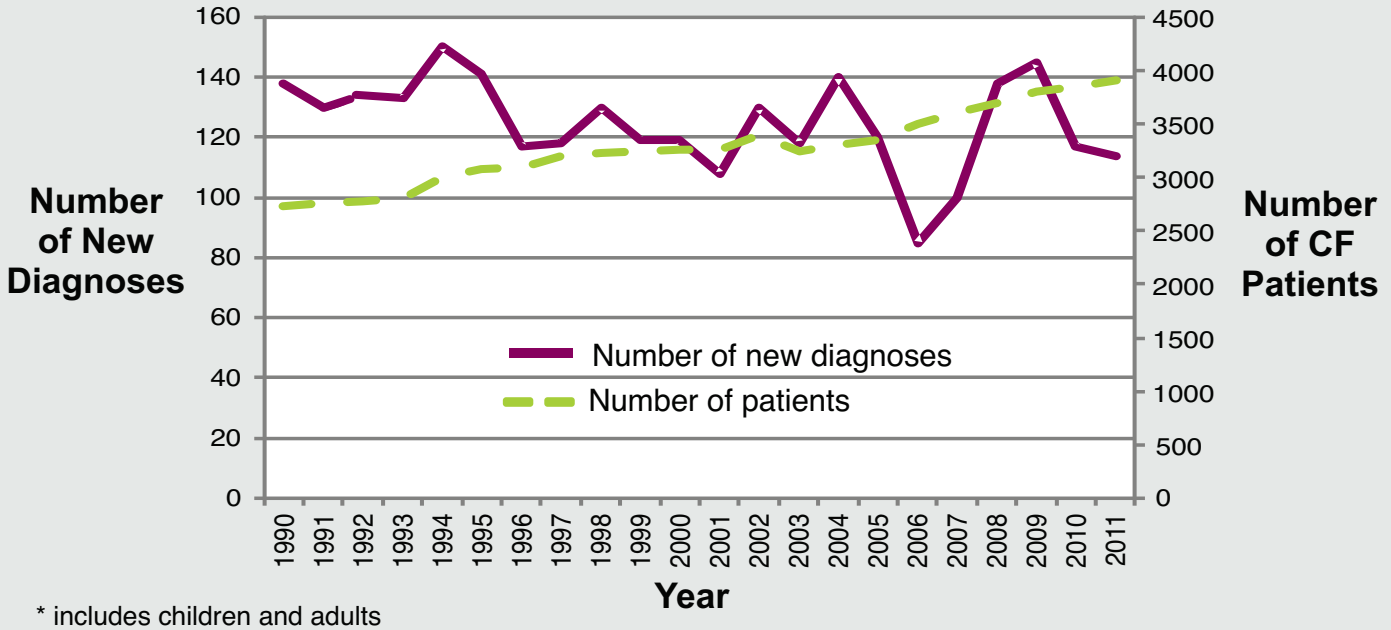
[1] Cystic Fibrosis Canada, About CF

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

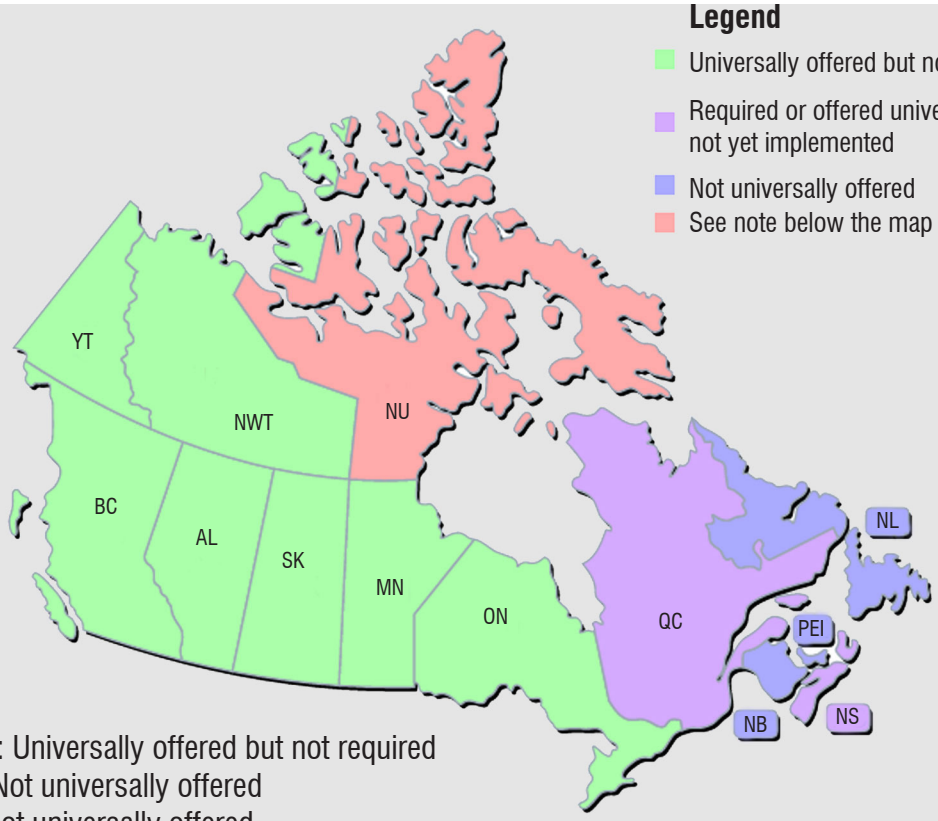


* includes children and adults

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

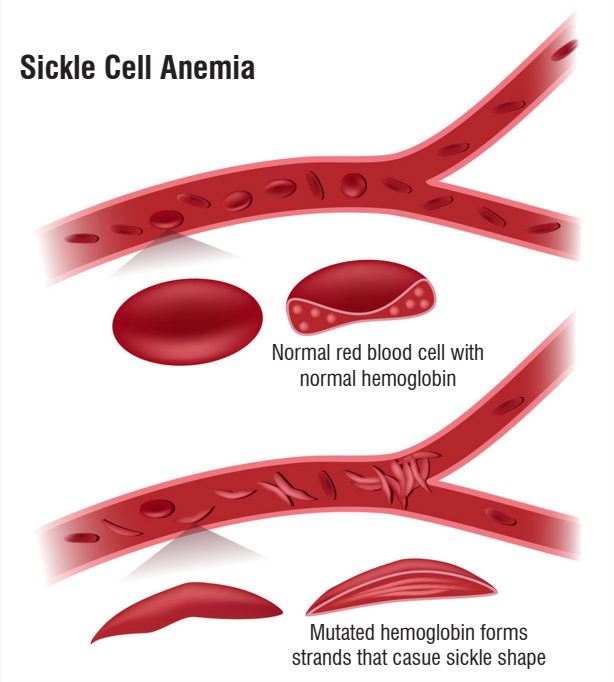


Image credit: Shutterstock

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.

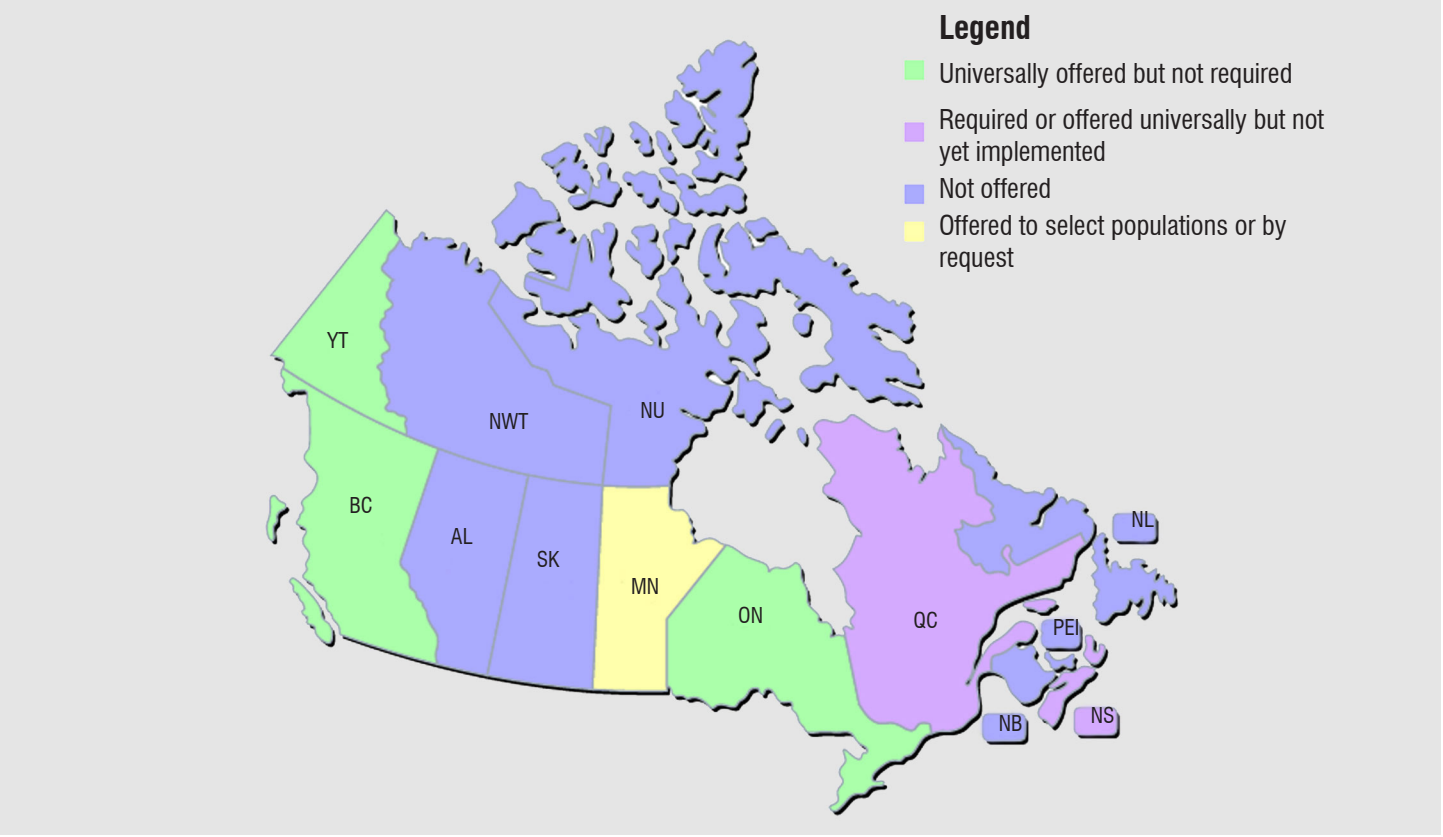
Sickle cell disease affects millions of people around the world. In Canada, there are about 2,000 people living with the disease. Up to 1 in every 2,500 babies born in Canada will have the disease.[1] Sickle cell disease is most common in families of African ancestry, but children of Middle Eastern,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

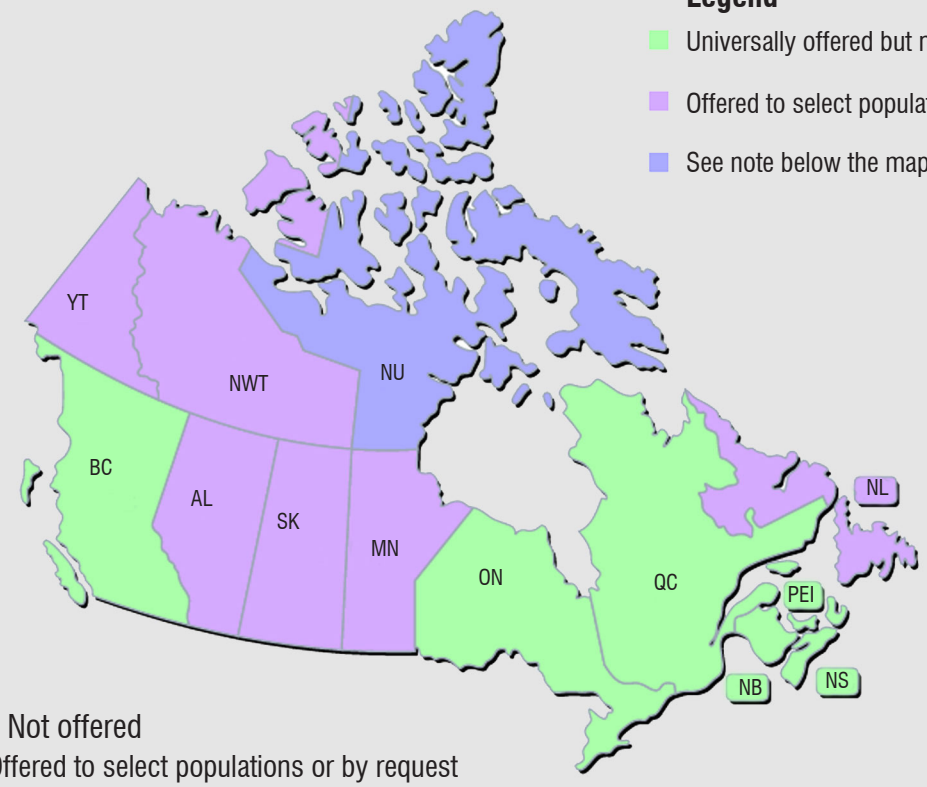
Implications

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