Canadian Agency for Drugs and Technologies in Health
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Newborn Screening for Disorders and Abnormalities in Canada

Environmental Scan

Context

The purpose of newborn screening for hereditary disorders is to identify neonates with treatable conditions. Early detection can prevent and/or minimize the potential for serious complications that can cause permanent damage or death.

Newborn screening started in Prince Edward Island in 1963. By 1970, most provinces had developed screening panels. In 1965, Ontario introduced universal screening for phenylketonuria, an inherited metabolic disease. The introduction of automated tandem mass spectrometry technology in the 1980s allowed the phenylketonuria blood spot to be used to screen for an additional 30 to 40 conditions.[1]

Many of the screened diseases are rare individually, but when considered together, are a major cause of pediatric morbidity and mortality. Some of the diseases are unfamiliar even to pediatricians, thus complicating diagnosis without screening.[2] Today, the most common disorders identified from newborn screening are congenital hypothyroidism, cystic fibrosis, medium-chain acyl-coenzime A dehydrogenase or MCAD deficiency, and phenylketonuria.[3]

In May 2011, the Centers for Disease Control and Prevention named newborn screening one of the top 10 great public health achievements of the last decade.[4]

Canada does not have a universal newborn screening panel for hereditary disorders. Each province and territory has its own newborn screening program and each program differs in the number of disorders screened, the rationale for screening, the technologies used for screening, legal structures and patient consent issues, governance and the use of advisory committees, treatment and follow-up processes, and coverage of disorders identified through screening.[5]

Objectives

The purpose of this report is to provide a general overview of newborn screening in Canada, including resources used and the retention period for samples. The following questions will be addressed:

- What hereditary disorders and abnormalities are newborns screened for?
- Where is newborn screening based in each jurisdiction?
- What technology is used for screening analysis?
- What is the specimen retention period for blood samples?

Findings

The findings of this environmental scan are not intended to provide a comprehensive review of the topic. Results are based on a limited literature search and communication with key informants from provinces and territories. This report is based on information gathered as of June 2011.

A national screening program does not exist in Canada. The federal government has no role in newborn screening beyond the licensing of some tests and other medical devices, and the regulation of foods, drugs and supplements for the treatment of rare and orphan diseases.

Health care and screening programs fall under provincial and territorial jurisdiction.

Information on provincial newborn screening panels is provided by the Canadian Organization for Rare Disorders, which periodically updates a status report, *Newborn Screening in Canada*. This report provides a comprehensive list of conditions, by province and territory, which are universally funded. The report can be found at: http://raredisorders.ca/documents/CanadaNBSstatusupdatedNov.112010.pdf

There is a wide spectrum in the number of disorders for which screening is offered, ranging from five disorders to 38, depending on the jurisdiction. All jurisdictions offer screening for congenital hypothyroidism and phenylketonuria. Saskatchewan is the only province in which it is a legal requirement to screen for both of these conditions. The legal enforcement of newborn screening has not been implemented in any other jurisdiction. In July 2010, the Canadian College of Medical Geneticists recommended that all provinces screen newborns for cystic fibrosis.[6] Currently, British Columbia, Alberta, Saskatchewan, Ontario, Yukon, and the Kitikmeot region of Nunavut screen for cystic fibrosis.

Blood spot samples are collected in hospitals; in the case of a home birth, a midwife or pediatrician will complete the test. Blood spots are collected from infants on a filter paper blood specimen card, most commonly using the heel-prick method. The specimen is taken ideally between two and three days after birth. Blood spots are then sent to laboratories within their jurisdiction. Most provinces have a single centre where blood spots are tested, but some provinces may have two or even three testing sites. Prince Edward Island and New Brunswick send their blood spots to the Izaak Walton Killam Health Centre in Halifax, Nova Scotia. The territories send their tests to sites in either British Columbia or Alberta. Information on the location of provincial newborn screening laboratories is provided in Table 1.

Table 1: Location of Newborn Screening Centres, 2011

JURISDICTION	LOCATION OF SCREENING CENTRE
British Columbia	• BC Prenatal Biochemistry Laboratory at Children's and Women's Health Centre of BC, Vancouver
Alberta	• Walter Mackenzie Health Sciences Centre, Edmonton • University of Calgary, Calgary Laboratory Services, and the Department of Pathology and Laboratory Medicine
Saskatchewan	• Saskatchewan Disease Control Laboratory, Regina
Manitoba	• Cadham Provincial Laboratory, Winnipeg
Ontario	• Children's Hospital of Eastern Ontario, Ottawa
Quebec	• CHU Sainte-Justine, Montreal • Sherbrooke • Quebec City
Prince Edward Island	• IWK Health Centre, Halifax, Nova Scotia
New Brunswick	• IWK Health Centre, Halifax, Nova Scotia
Nova Scotia	• IWK Health Centre, Halifax
Newfoundland and Labrador	• Health Sciences Centre, St. John's
Yukon	• BC Prenatal Biochemistry Laboratory at Children's and Women's Health Centre of BC, Vancouver
Northwest Territories	• Walter Mackenzie Health Sciences Centre, Edmonton, Alberta
Nunavut	• Walter Mackenzie Health Sciences Centre, Edmonton

Tandem mass spectrometry is used in hospitals in each province to detect newborn abnormalities and disorders. This technology is not appropriate for the screening of some conditions, including cystic fibrosis and congenital hypothyroidism.

Other technologies used to detect newborn abnormalities include immunoassays using fluorescent detection and spectrophotometric techniques.

Currently, no regulatory policy in Canada governs the retention and secondary use of blood spot samples. The retention period for blood spot samples varies broadly across jurisdictions,[7] ranging from two to 28 years. The storage of blood samples has been the cause of legal action. A class action suit was filed in the Supreme Court of British Columbia that challenged the retention and use of newborn blood samples without explicit parental consent.

Today, after initial tests are complete, the cards containing the blood samples are stored for further clinical research purposes. Concerned parents and privacy advocates have objected to non-consensual secondary use, particularly since blood samples contain personal genetic details. The main concern is that the longer samples are stored, the greater the risk that unknown third parties may access them. The proposed British Columbia class action (*Doherty v. Provincial Health Services Authority*) notes that "potential users" of the samples "include law enforcement personnel...and health insurers," and that "expansion of the range of information that can be extracted from the blood is reasonably foreseeable."

In a recent policy change, parents in British Columbia now have the option of requesting that their children's blood sample cards be destroyed following the completion of the tests. In other provinces, such requests may be handled on a case-by-case basis. The Ontario program, for example, does not have a formal policy on sample card destruction directives, but suggests that sample cards should be retained for at least five years for any necessary follow-up testing.[8] The provincial retention period for specimens is presented in Table 2.

Table 2: Retention of Specimens, 2011

Jurisdiction	Specimen Retention (in years)
British Columbia and Yukon	10
Alberta	7
Saskatchewan	21
Manitoba	28 (10 years + age of majority)
Ontario	5
Quebec	Not available
Maritimes	5
Newfoundland and Labrador	2

The American College of Medical Genetics has a Position Statement on the Importance of Residual Newborn Screening Dried Blood Spots.[9] The College states that dried blood spots have additional value beyond screening. For example, they are essential for quality improvement of newborn screening tests and are critical in the development of new screening tests. No publicly accessible Position Statement was identified by the Canadian College of Medical Geneticists.

Conclusion

Newborn screening is universally available in all Canadian jurisdictions. These programs are funded by the provincial/territorial health insurance plans. While there are some similarities between programs, specifically regarding testing methods, jurisdictional newborn screening programs differ in the delivery of the service, the number of disorders screened, and the retention period for blood samples.

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