Alberta Newborn Metabolic Screening Program

POLICY DOCUMENT

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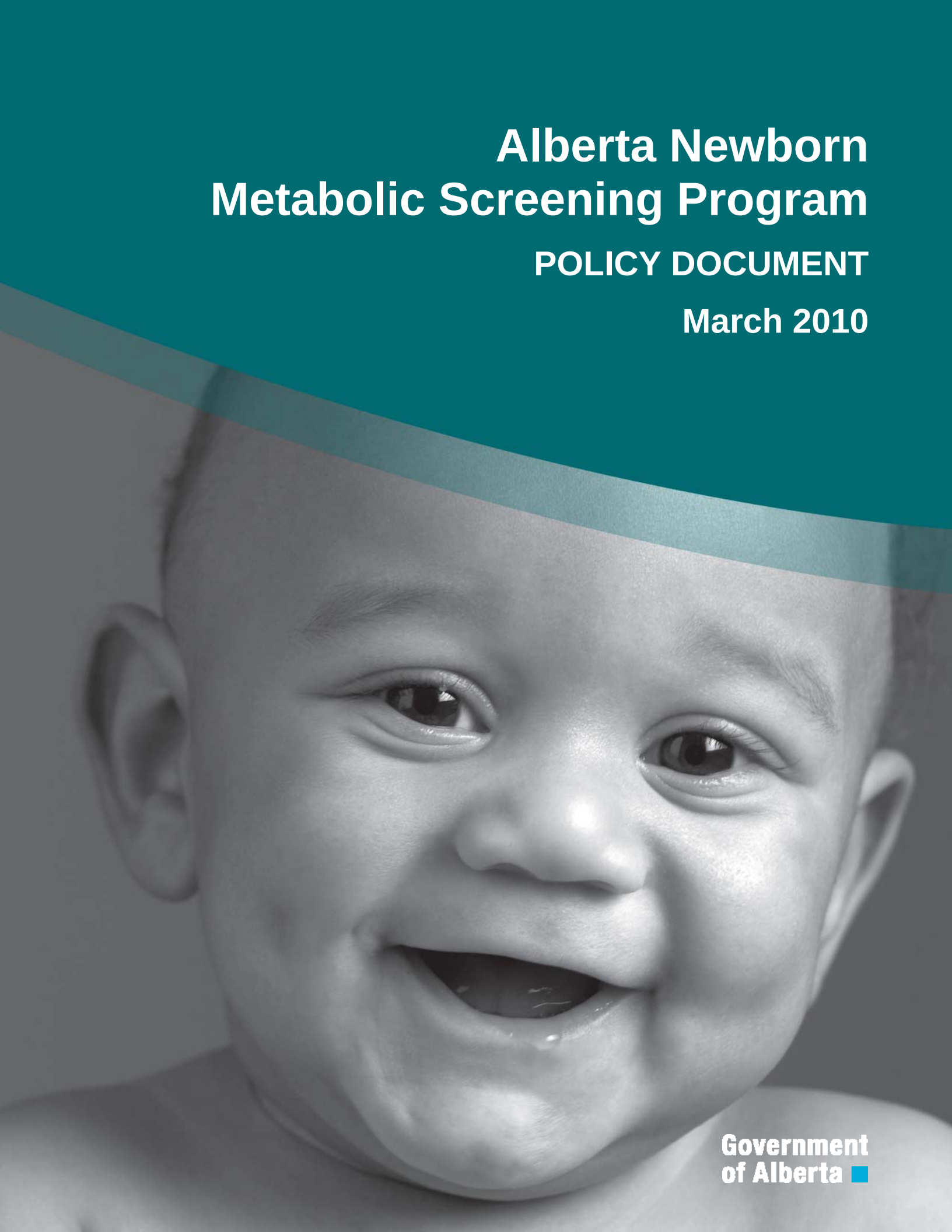

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Table of Revisions

Version	Revision Date	Revision History
1.0	2010-05-31	Document released.
1.1	2011-01-10	Program Scope revised, Appendix B revised, minor grammar corrections in document body.

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Introduction

Each year, a small number of Alberta babies are born with metabolic disorders that can lead to severe physical disabilities, developmental delay, other health problems or even death. The purpose of the Newborn Metabolic Screening (NMS) program is to identify and treat infants for these disorders before the onset of symptoms. Newborn screening is time sensitive. Babies affected with some of the screened disorders will start to become ill and may suffer irreversible damage soon after birth. Early effective treatment can better manage these disorders and decrease morbidity and mortality.

Purpose

The purpose of this document is to set out:

- NMS program policies; and
- roles and responsibilities for implementing the NMS program.

Background

The newborn screen is carried out on a blood sample collected from the baby's heel on filter paper between 24 hours and 72 hours of age. The newborn screen identifies babies who have a high risk of having one of the metabolic disorders included in the testing menu. The condition is confirmed by clinical examination and diagnostic testing.

Newborn metabolic screening in Alberta started in 1967, with the introduction of screening for phenylketonuria (PKU). Screening for congenital hypothyroidism (CH) was added in 1977 and screening for biotinidase deficiency was added in 1990. On April 1, 2007, Alberta expanded its NMS program to screen for 17 disorders, including 14 metabolic disorders, two endocrine disorders and cystic fibrosis (CF). (See Appendix A for list of disorders). While the list of disorders being screened for has expanded beyond metabolic disorders, the program will retain its original name.

A web-based NMS tracking system (the 'NMS Application') enables Alberta Health Services to track all registered births and newborn metabolic screens, and to aid follow up of those babies requiring repeat screening.

Program Goals

- Morbidity and mortality of Alberta infants with the screened disorders is minimized through timely and effective screening to allow the early diagnosis and treatment of affected infants.
- All babies born in Alberta have timely access to newborn metabolic screening as an integral component of their health care. The program goal is that all babies have an initial screen reported on or before the 10th day of age.

- Parents, health professionals and the public are informed about the NMS program.

Program Scope

Newborn metabolic screening is a part of the health care provided to all infants born in Alberta. The population of infants born in Alberta is the primary priority for this program.[1]

The NMS program begins immediately after registration of an Alberta live birth in the Alberta Health and Wellness Person Directory application and ends with a negative screen result or referral to diagnostic and treatment services when a positive screen is identified. Screening is not diagnostic – positive results are referred for further testing to rule out or confirm a disorder. The outcomes of diagnostic testing are collected as part of quality assurance for the program.

While newborn screening is an important tool in improving health outcomes, it is neither comprehensive nor is it sufficient in all cases. It does not replace the need for parents and health care providers to pay close attention to an infant's health. Specifically, it does not replace the responsibilities or professional judgment of health care providers and, in particular, primary care providers. For infants where there is an elevated risk of a screened disorder due to family history or children exhibiting symptoms of a disorder, it is the responsibility of the attending primary care provider to arrange any screens, tests or follow-up that, in the primary care provider's opinion, is required.

[1] The standards in this document may vary to accommodate babies born outside of Alberta and not registered as newborns in Person Directory, but screened in Alberta.

Roles and Responsibilities

A number of organizations have responsibility for implementing the NMS program.

Alberta Health and Wellness (AHW):

- sets the policies and standards for the program;
- oversees Alberta Health Services' NMS program delivery plan;
- determines what conditions are screened for;
- facilitates co-ordination with stakeholders;
- monitors compliance with NMS policy;
- sets monitoring and reporting requirements for Alberta Health Services;
- appoints and maintains an advisory structure as required to provide ongoing advice to the ministry;
- designates an AHW NMS program contact;
- communicates at a provincial and federal level on matters of newborn screening policy;
- may develop a surveillance plan;
- maintains the Person Directory and NMS Applications; and
- provides direction to stakeholders as needed.

Alberta Health Services (AHS):

- develops and implements the NMS program delivery plan, which includes:
    - development and dissemination of provincial NMS procedures for implementing current AHW policy;
    - development of training materials and training for program delivery;
    - quality assurance activities and planning;
    - a business continuity plan which identifies and addresses key organizational positions and infrastructure required for program delivery;
    - coordinated delivery of NMS program components;

  - development of a communication plan;
  - protocols to provide timely and effective assessment, diagnosis and treatment of screen positive infants;
  - development of policies and procedures on sample storage and retention; and
  - processes to develop, revise, procure and distribute requisitions.

- designates a senior executive with accountability for the NMS program delivery plan;
- monitors and reports on program performance as required by AHW;
- properly registers all infants born in Alberta in the Person Directory application;
- maintains capacity/expertise/infrastructure for NMS program delivery and follow up of infants who screen positive at all times;
- collects newborn screening samples and completes the NMS requisition in accordance with AHW policies;
- provides parents with sufficient information to understand the purpose of the screen and the reasons for it;
- participates in informational sessions with key stakeholders;
- develops and prepares operational informational materials and provides input into program informational materials; and
- provides data for surveillance purposes as required by AHW.

<u>AHS Newborn Metabolic Screening Laboratory/AHS Molecular Diagnostic Laboratory:</u>

- tests all samples in accordance with the College of Physicians and Surgeons Laboratory Accreditation Standards;
- reports the results in accordance with AHW policy; and
- submits test results to the NMS Application according to AHW data submission requirements.

AHS Metabolic, Cystic Fibrosis and Pediatric Endocrinology Clinics:

- accept referrals and facilitate assessment and follow up in a timely manner;
- participate with the NMS lab, as required, to determine cut-off points for screening testing results for referral; and
- notify the NMS lab of the final outcome of diagnostic studies on all patients referred from the NMS lab.

Primary Care Providers:

- if participating in sample collection, collect the sample and complete the NMS requisition in accordance with AHW policies;
- provide parents with sufficient information to understand the purpose of the screen and the reasons for it;
- act promptly on positive screening results by contacting the specialty clinics (such as cystic fibrosis clinic, metabolic clinic) and families; and
- notify the NMS lab of the final outcome of diagnostic studies on patients referred from the NMS lab.

Definitions

For the purpose of this document, the following definitions have been used:

Business days	Monday through Friday, except for statutory holidays.
Days	Refers to calendar days.
Incident	An act or failure to act that causes, or that may cause harm to a newborn.
Newborn	Babies born in Alberta, 28 days or less in age and registered in the Person Directory as “add newborn”.
NMS Application	A secure, web-based application that reconciles registered births and newborn metabolic screen results and generates alerts which assist in identifying and tracking all infants requiring follow up.

NMS Contact	NMS Application user who reports to NMS Manager.
NMS Manager	NMS Application user responsible for managing the NMS Program in a designated geographical area. This includes monitoring the NMS Contacts as well as area performance.
NMS Program	AHS' NMS program delivery plan as approved by AHW.
Electronic Health Record (EHR)	A secure, web-based application that provides authorized health professionals with confidential access to essential demographic, medication and allergy profile, and lab test results information.
Person Directory (PD) application	A secure, web-enabled application that delivers person-identifiable demographic and eligibility information to authorized health service providers.
Policy	General principles that set the direction for the NMS program and by which the organizations responsible for the NMS program are guided in the delivery of this program.
Primary Care Provider	Family physician, pediatrician, nurse practitioner or midwife.
Specialty Clinics	Refers to the metabolic, cystic fibrosis and pediatric endocrine clinics operated by AHS.

Policies

Accountability	
1. Clear accountability and lines of responsibility are a requirement for effective program delivery.	1.1 AHS must implement and deliver the NMS program.
	1.2 The CEO of AHS shall be accountable for whether the NMS Program operates in accordance with current AHW policy and standards.
	1.3 AHS must designate a senior executive with responsibility for the NMS program delivery plan and adherence to current AHW policy and standards.

1.	Continued	1.4 AHS must designate NMS managers and NMS contacts with responsibility for monitoring and reporting on the NMS program, in sufficient numbers to operate the program in adherence to current AHW policy and standards.

Program Delivery Timeline		
2.	The goal of the program is that all babies have an initial screen reported on or before the 10th day of age (except for DNA testing of CF results).	2.1 The expectation is that AHS will align its processes so that initial screens are reported wherever reasonably possible on or before the 10th day of age.
		2.2 Guidelines provided within this document are benchmarks for processes in the screening process and shall not supersede the overall reporting standard of 10 days.
		2.3 AHS must evaluate all aspects of program delivery and whenever possible reduce delays and strive to minimize the period of time between birth and reporting of screening result.
		2.4 AHS must develop an emergency preparedness plan and maintain delivery of NMS program services during emergencies and infectious disease events.

Procedure and Operational Policies		
3.	Procedures and operational policies related to program activity must be developed, implemented provincially and maintained to reflect current AHW policy.	3.1 Operational policies, procedures, plans, and training and informational materials related to NMS program delivery must be submitted to AHW on request and AHW may request modifications to these documents.
		3.2 AHW must be notified of any significant operational changes to the NMS Program in a timely manner.

Access		
4.	Newborn Metabolic Screening is part of the health care services provided to infants born in Alberta.	4.1 AHS must make all reasonable efforts to screen all infants born in Alberta.
		4.2 AHS must provide training to staff registering newborns in the Person Directory application, obtaining the screening samples and accessing the NMS Application.
		4.3 AHS must register infants born in Alberta in the Person Directory application through the “add newborn” function within 24 hours of birth.
5.	Newborn metabolic screening is part of the standard of care that every baby born in Alberta receives. Parents/guardians must be informed about the nature and purpose of newborn metabolic screening.	5.1 The health professional taking the sample for screening or the primary care provider who is requisitioning the sample must inform the parent of the reason for the screen and provide information about the program before the sample is collected.

Refusals	
6. If parents/guardians decline to have the newborn screen sample collected it is important that they are informed of the purpose of the test and the possible implications of refusing the screen.	6.1 Parents/guardians who decline the screening must be informed by the health professional arranging sample collection or the primary care provider who is treating the infant about the possible implications of the infant not being screened. Refusal of screening must be adequately documented by the health professional in accordance with direction from AHS' legal counsel.
7. Newborn metabolic screening applies to all disorders in the screening list; there is no option for selective screening.	7.1 Parents/guardians who do not wish their child to be screened for all the disorders are therefore refusing the screen and must be immediately referred, by the health professional providing care, to their physician for follow up and the parents' decision must be documented.
	7.2 Parents/guardians who accept screening and request additional testing for disorders not on the list should have the newborn's sample collected as per protocol and referred to their physician for any additional screening.
8. Screening will not be provided in cases where a physician advises that screening should not be done for a clinical reason.	8.1 It is the responsibility of AHS to obtain legal advice on how to respond to a physician who advises against screening and to document physician refusal of screening.

Sample Collection and Delivery	
9. If infant is born in Alberta, AHS is responsible for obtaining the sample for newborn metabolic screening and for transporting samples to the NMS lab within the specified timeframe.	9.1 AHS must collect initial samples from infants between 24 hours and 72 hours of age and as close to the 24 hours as reasonably possible.
	9.2 AHS must collect a repeat screen between three and six weeks of age for all infants who are in the neonatal intensive care unit (NICU) for three weeks or longer.
	9.3 Samples must be taken according to the instructions provided by the NMS laboratory.
	9.4 The health professional taking the sample must fill out the NMS requisition completely and accurately.
	9.5 AHS shall create, monitor and manage a province-wide system to deliver samples to the NMS lab.
	9.6 AHS must deliver samples to the NMS lab within three days of sample collection.

Alert Management	
10. AHS is responsible for monitoring alerts in the NMS Application and ensuring appropriate actions are performed in accordance with AHW policies and standards.	10.1 NMS contacts must check the NMS Application for alerts and perform activities related to alert management every business day.
	10.2 When an alert is posted, AHS must collect and submit a sample within 4 days of the alert posting (except for increased tyrosine).
	10.3 In the case of increased tyrosine, AHS must collect and submit a sample between 30 and 42 days of age.

10. Continued	10.4	AHS must implement a process for contacting families for screens and efforts to contact the family must be documented in the NMS Application.
	10.5	AHS must implement a process for determining when it is appropriate to manually close alerts in the NMS Application (when efforts to collect the sample have been unsuccessful) and explanations for closing alerts must be documented in the NMS Application.
	10.6	If the newborn has moved out of province, AHS shall notify a person responsible for NMS in the jurisdiction where the infant resides of the status of screening and document notification in the NMS Application.

Sample Testing		
11. AHS must designate a single laboratory as the provincial NMS laboratory.	11.1	The NMS laboratory is responsible for timely testing of all initial and repeat screens.
	11.2	The NMS lab must be fully operational on all business days. It must not be closed for more than two consecutive days.
	11.3	The NMS laboratory must accurately record the date and time when a sample arrives in the NMS laboratory.
	11.4	The NMS laboratory must enter a sample in the laboratory information system whenever possible on the day received and in any event within two days of arrival.
	11.5	The NMS laboratory must analyze samples whenever possible on the day received and in any event within two days of arrival.

11. Continued	11.6 The NMS laboratory must check the NMS Application for alerts and perform activities related to alert management every business day.
	11.7 Laboratory policies related to retention, security, access to, quality assurance and reporting of NMS samples must be submitted to AHW on request and AHW may request modifications to these documents.
12. AHS must designate a single laboratory as the NMS Molecular Diagnostic Laboratory.	12.1 The Molecular Diagnostic Laboratory is responsible for the DNA testing component of CF screening.

Research	
13. The sample is collected only for the purpose of NMS screening for the newborn.	13.1 All research requests regarding access to NMS samples must be made in accordance with the requirements of the *Health Information Act*.

Reporting of Results	
14. NMS lab results must be reported to the: • primary care provider as documented on the requisition, • the birth facility, • the NMS Application, and the Electronic Health Record (EHR).	14.1 The NMS lab must issue an alert or have test results posted within four days of receipt of the sample (except for DNA testing for CF).
	14.2 The molecular diagnostic lab must report CF DNA results on referred specimens to the NMS lab. The NMS lab must issue an alert or have test results posted for CF DNA results within 21 days of receipt of the sample.
	14.3 AHS must report NMS laboratory data electronically to the NMS Application in a format acceptable to AHW, and the delivery of the electronic messages must be monitored and any interruption to delivery identified, reported to AHW and resolved in a timely manner.

15. All children with abnormal screen results must receive timely and appropriate referral for follow up and treatment.	15.1 AHS must establish protocols for referral and follow up for each of the disorders screened for in consultation with specialty clinics and AHW.
	15.2 The NMS lab must report abnormal test results to primary care providers and specialty clinics in accordance with the AHS protocols for referral and follow up.

Diagnosis and Treatment	
16. It is the responsibility of primary care providers and specialty clinics to follow up with families and provide timely assessment, diagnosis and treatment.	16.1 Primary care providers receiving notification of abnormal test results must follow up with the family and/or specialty clinic as appropriate or immediately notify the NMS lab if unable to contact the family.
	16.2 Specialty clinics must contact the family upon notification by the NMS lab that there is no primary care provider, or one cannot be contacted, or upon the request of the primary care provider.
	16.3 Specialty clinics and physicians must provide diagnostic results (when available) to the NMS lab for quality assurance purposes.

Incident Reporting and Investigation	
17. The reporting and investigation of incidents must identify potential risks, actions required to prevent similar occurrences and opportunities for quality improvement.	17.1 Any individual who knows or suspects of an incident must report the incident to the Alberta Health and Wellness NMS program contact in a timely manner.
	17.2 On request from AHW, the designated AHS senior executive responsible for NMS program delivery must investigate incidents promptly and report outcome, recommendations and steps taken to AHW.
	17.3 AHW may perform its own investigation of incidents at its discretion.

Appendix A: List of Disorders

Effective April 1, 2007

Amino acid disorders
Citrullinemia (CIT)
Maple syrup urine disease (MSUD)
Phenylketonuria (PKU)

Organic acid disorders
Glutaric acidemia type 1 (GA1)
3-Hydroxy-3-methylglutaryl-CoA lyase deficiency (HMG)
Isovaleric acidemia (IVA)
Methymalonic acidemia (MMA)
Propionic acidemia (PA)

Fatty acid oxidation disorders
Carnitine uptake defect (CUD)
Long chain hydoxyacyl-CoA dehydrogenase deficiency (LCHAD)
Medium chain acyl-CoA dehydrogenase deficiency (MCAD)
Trifunctional protein deficiency (TFP)
Very long chain acyl-CoA dehydrogenase deficiency (VLCAD)

Endocrine Disorders
Congenital adrenal hyperplasia (CAH)
Congenital hypothyroidism (CH)

Other disorders
Biotinidase deficiency (BIOT)

Cystic Fibrosis

Appendix B: NMS Program Manuals and Brochures

Alberta's Newborn Metabolic Screening Program, A Healthy Beginning for Your Baby

NMS disorder fact sheets

NMS Application Manual

Registration User Guide – Person Directory