



Alberta Perinatal Connection

Alberta's Newborn Metabolic Screening Program

In this issue:

From the APHP Director	1
Connections	1
Connected through Education	2
Connected Through Information	3
Connected Through Quality	4
Contact Information	4

Alberta Perinatal Connections is published twice yearly for the perinatal Professional Community in Alberta by the Alberta Perinatal Health Program. This publication is edited by Lesly Deuchar, who can be contacted at Lesly.Deuchar@capitalhealth.ca

The Alberta Perinatal Health Program would like to acknowledge Irene Mazurenko from Alberta Health and Wellness for her time in ensuring the accuracy of the content of this publication... Thank-you, Irene.

For more information on the NMS program in Alberta, contact Irene Mazurenko, Project Manager, Alberta Health and Wellness, at 780-427-3488 or Irene.Mazurenko@gov.ab.ca

From the APHP Director

Continuing with the theme of 'Connections' among the Perinatal Professionals and their communities throughout Alberta, we welcome you to this second edition of Alberta Perinatal Connections newsletter. Emphasizing the value and strength of connections between community care and acute care; this issue will focus on the recently expanded Newborn Metabolic Screening (NMS) program in Alberta.

The strength of the connections that exist between both the hospital and community settings is determined by the

professionals working in each of these settings. Our shared goal of seamless service delivery to the families we care for is what fosters these connections.

Please enjoy reading this newsletter and feel empowered to reach out to other perinatal professionals in your community to further strengthen the Alberta Perinatal Connections!



Newborn Metabolic Screening in Alberta

In 2007, Alberta increased the number of tests performed through the NMS program to include screening for 17 disorders. This screening program is one of the most comprehensive in Canada and relies upon efficient processes between community and acute care services to ensure that all infants in the province are screened and that all screening results are reported back to the primary care provider in a timely fashion.

Routine screening for inborn errors of metabolism that can be diagnosed and treated before the onset of irreversible damage has been the practice in Alberta since 1967, starting with phenylketonuria. Congenital hypothyroidism was added in 1977 and biotinidase deficiency in 1990.

Just over 40 years after the first screening program was implemented,

newborn metabolic screening now includes: three Amino Acid Disorders, five Organic Acid Disorders, five Fatty Acid Disorders, two Endocrine Disorders as well as Biotinidase Deficiency and Cystic Fibrosis.

Registered newborns in Alberta are tracked in an NMS application to ensure they are offered screening. Tracking the screening of infants ensures that follow-up is completed as well.





Screening through the NMS program is not mandatory. It is offered for all infants because it allows diagnosis and treatment to occur before irreversible damage occurs.

Parents and guardians need to know that newborn metabolic screening is part of the health care provided to all newborns in Alberta.

The screening test is performed on a blood sample collected from each baby between 24 hours and five days of age.

ALL NMS samples collected in Alberta are sent to the Capital Health Newborn Metabolic Screening laboratory for testing.

Follow-up of positive NMS screens is done by primary care providers and specialty clinics (cystic fibrosis, metabolic, pediatric endocrinology).

Newborns who are in the hospital for three weeks or longer need to have a repeat screen between three and six weeks of age.

The parent brochure: *Alberta's Newborn Metabolic Screening Program, A healthy beginning for your baby* is available through your local public health office or at www.health.alberta.ca/public/NMS_PublicBrochure.pdf.

A brochure for health professionals and fact sheets on each of the conditions can be found at www.health.alberta.ca/professionals/health-resources.html.

Connected Through Education

Tests Included in the Newborn Metabolic Screening Program

Amino Acid Disorders		
Citrullinemia	CIT	The inability to break-down certain amino acids allows toxic levels to develop. Treatment is through diet.
Maple syrup urine disease	MSUD	
Phenylketonuria	PKU	
Organic Acid Disorders		
Glutaric aciduria type 1	GA1	Accumulating organic acids result from an enzyme defect affecting metabolism of certain amino acids. Affected infants become ill in the first days of life. Treatment is through diet.
3-Hydroxy-3-methyl glutaric acidemia	HMG	
Isovaleric acidemia	IVA	
Methylmalonic acidemia	MMA	
Propionic acidemia	PA	
Fatty Acid Oxidation Disorders		
Carnitine uptake defect	CUD	Absence of enzymes involved in fatty acid breakdown, with subsequent accumulation of fatty acids and vital organ damage ensues. Treatment is through diet... fasting is avoided.
Long chain hydroxyacyl-CoA dehydrogenase deficiency	LCHAD	
Medium chain acyl-CoA dehydrogenase deficiency	MCAD	
Trifunctional protein deficiency	TPF	
Very long chain acyl-CoA dehydrogenase deficiency	VLCAD	
Other Metabolic Disorders		
Biotinidase deficiency	BIOT	Lack of enzyme prevents biotin recycling. Treated with supplementation of biotin.
Endocrine Disorders		
Congenital adrenal hyperplasia	CAH	Absent hormones affect growth and development. Treated with hormone replacements.
Congenital hypothyroidism	CH	
Other		
Cystic fibrosis	CF	An inherited disease – thickened secretions affect pulmonary and digestive systems. Treated with enzyme replacements.

Healthcare professionals who work in settings where early contact with expectant or new parents occurs need to be able to discuss the NMS program. Because the screening program has the potential to prevent serious

health problems, or save a baby's life, it is important that parents have all the necessary information to make the best choice for their baby.



Connected Through Information

Births that Occurred in Alberta Hospitals in 2006

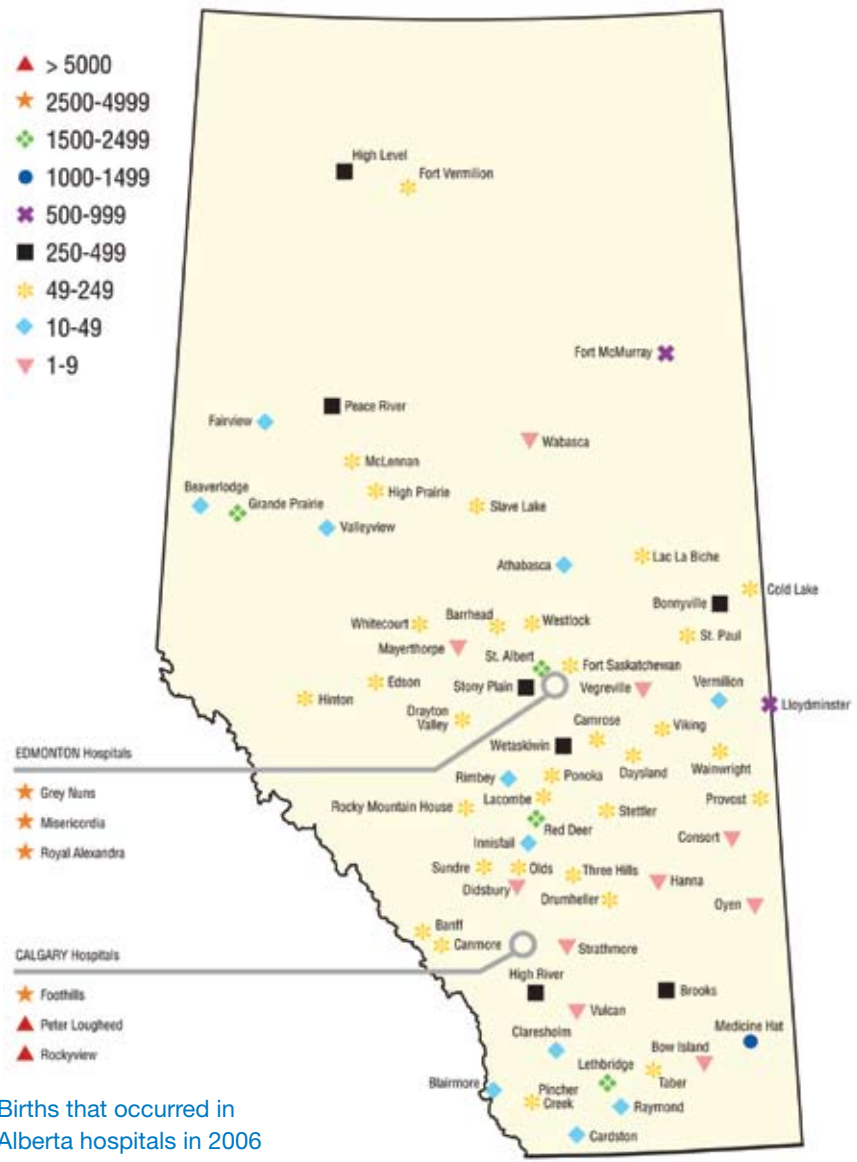
Each health region in Alberta has a designated manager for the NMS program as well as processes in place for obtaining screens on newborns.

Every newborn in Alberta must be registered in the Alberta Health and Wellness Person Directory within 24 hours of the birth. Newborns are registered as a 'NEWBORN' and are registered in the 'Person Directory' by the health region in which they were born.

At registration, each newborn is issued a Unique Lifetime Identifier (ULI) which facilitates tracking to ensure that all are offered the screening test. If they are not registered as newborns they cannot be tracked in the NMS application.

Newborns that are registered more than one time (perhaps due to patient transfers), or under more than one name may not be tracked as efficiently as correctly registered newborns.

Alberta is the first Canadian NMS program to screen NEWBORNS for Cystic Fibrosis.



In 2007, Alberta Health and Wellness reports 48,647 live births in Alberta, which represents a 7% increase over 2006. The screening rate for newborns in Alberta continues to be greater than 99%, despite this marked increase in number of births.

Even with this increase in the number of births, the follow-up processes for the screening on these newborns continues to be strong. Working through connections that exist in the regions and between the regions, infants are tracked to ensure they receive screening, as well as any repeat screens and follow-up required.

