NEWS RELEASE

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Ministry of Healthy Living and Sport
Provincial Health Services Authority

B.C. ROLLS OUT EXPANDED NEWBORN SCREENING PROGRAM

VANCOUVER – Newborns across British Columbia are now being screened for more treatable metabolic diseases and other medical conditions, resulting in early identification and treatment.

The BC Newborn Screening Program, a service of the Provincial Health Services Authority (PHSA), has expanded screening from six to 18 disorders, using the same simple blood sample already collected shortly after birth. Over the coming year, the program will screen about 40,000 newborns in B.C. and will likely identify about 40 babies with one of these 18 treatable conditions. In October 2010, a 19th disorder (congenital adrenal hyperplasia) will be added to the list of screened conditions for newborns. This follows through on a commitment made by Premier Campbell in July 2008 for the additional screening tests.

"This program provides newborn babies in British Columbia with their best chance for a healthy life, right from the start," said Ida Chong, Minister of Healthy Living and Sport. "With specialized treatment, newborns identified with these disorders can avoid serious complications, such as development problems, liver malfunction, brain damage, respiratory problems and even the risk of sudden death."

B.C. began newborn screening in 1964 with provincewide screening for one disorder – phenylketonuria (PKU). Prior to this current expansion, the BC Newborn Screening Program screened for six treatable disorders including PKU, congenital hypothyroidism, galactosemia, medium-chain acyl-CoA dehydrogenase deficiency (MCADD), glutaric aciduria Type I (GAI), and long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency (LCHAD).

"Screening for these 18 treatable disorders improves health outcomes for those affected children. These disorders were chosen after a careful review of the medical literature and best practices," said Dr. Hilary Vallance, medical director of the BC Newborn Screening Program, director of the biochemical genetics laboratory at BC Children's Hospital and BC Women's Hospital & Health Centre, and chair of the newborn screening advisory committee. "There are no additional steps for families or health-care providers, as the testing utilizes the same few drops of blood taken from the heel of a newborn that has been collected from babies born in B.C. for more than four decades."

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"The newborn screening program is an excellent example of the kind of innovation we are striving to develop throughout our health care system," said Wynne Powell, board chair for the PHSA. "New and improved technologies enable earlier identification and treatment of patients, which means our health dollars are used more effectively and efficiently to better support early treatment and help avoid long-term complications that can affect quality of life and be costly to our health system."

Annual operating costs for the expanded BC Newborn Screening program are provided by the PHSA and are approximately $2 million – representing an investment of about $50 for each baby born in B.C. each year.

With the expansion of its newborn screening program, B.C. becomes one of the first jurisdictions in Canada to implement second-tier testing for selected disorders. The second-tier test is currently used to confirm initial newborn test results indicating cystic fibrosis. By late 2010, second-tier testing will also be used to confirm abnormal results for six other conditions.

"I am certain that my 28-year-old daughter Kim would be in better health today had she been diagnosed with cystic fibrosis at birth," said Chris Black, past president of the Canadian Cystic Fibrosis Foundation. "When Kim was born, we endured more than a year of illness and uncertainty before she was diagnosed. I'm thrilled that babies born with cystic fibrosis in B.C. will have a better start in life than Kim did."

The BC Newborn Screening Program, a service of the PHSA, is a collaboration of BC Women's Hospital & Health Centre, BC Children's Hospital and the BC Perinatal Health Program, all agencies of the PHSA.

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A backgrounder follows, with a list of the additional disorders being screened for via the expanded BC Newborn Screening Program.

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BACKGROUNDER

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BC NEWBORN SCREENING PROGRAM

Below is a list of treatable disorders being screened for among newborns in British Columbia since expansion of the newborn screening program in November 2009. Screening is expected to identify about 40 newborns with these disorders each year. Also included in the list is congenital adrenal hyperplasia, a condition that will be added to the newborn screening list in October 2010.

1. Congenital hypothyroidism	A condition in which a baby cannot make enough thyroid hormone. Without it, delayed growth and brain damage occur. With early detection and thyroid hormone treatment, these children have normal growth and intelligence. One baby in 3,500 is born with hypothyroidism.
2. Galactosemia	A condition that affects a person's ability to process the sugar galactose, which, if untreated, can lead to liver, brain, and eye damage. These problems are prevented with a special diet.
3. Phenylketonuria (PKU)	A baby with PKU is missing an enzyme that is needed to process the essential amino acid phenylalanine, found in certain foods. Without treatment, phenylalanine builds up in the baby's blood and causes mental retardation. With early diagnosis and dietary treatment, mental retardation is prevented.
4. Glutaric Aciduria Type I (GA-I)	A baby with GA-I is missing an enzyme used in the breakdown of several amino acids. Babies may develop normally early on but are at risk of metabolic crisis that can lead to brain damage, seizures, and cerebral palsy-like symptoms. With early diagnosis and treatment, brain damage may be prevented.
5. Medium-chain Acyl-CoA dehydrogenase deficiency (MCAD)	A baby with MCAD may have problems using fats stored in their body for energy. The baby is healthy when eating well. If the baby has a cold or flu, he or she may not be able to use the stored fatty acids as energy. There is a risk of sudden unexpected death (similar to SIDS), which can be prevented by using a special diet and not fasting.
6. Long-chain Hydroxyacyl-CoA dehydrogenase deficiency (LCHAD)	A condition that prevents the body from converting certain fats into a usable energy source. LCHAD deficiency can present in many ways, such as low blood sugar, sudden unexpected death, low muscle tone, or problems with the function of the heart. Treatment with avoidance of fasting and a special diet can improve health outcomes.

Expanded Screening Panel	
7. Very-long chain AcylCoA dehydrogenase deficiency (VLCAD)	Similar to LCHAD, this is a condition that prevents the body from converting certain fats into a usable energy source. Low blood sugar and muscle symptoms can be prevented with dietary treatment and avoidance of fasting.
8. Citrullinemia	A disorder that causes ammonia and other toxic substances to accumulate in the blood. A build-up of ammonia can cause brain damage and can be life-threatening. Treatment with a special diet can improve health outcomes.
9. Tyrosinemia I	A missing enzyme in the breakdown of the amino acid tyrosine leads to liver and kidney damage. The disorder is treatable with medication and, in some cases, liver transplantation.
10. Homocystinuria (including second-tier testing)	A missing enzyme in the processing of the amino acid, homocysteine. The resultant high homocysteine levels in blood increase the risk of stroke. Early detection and treatment prevents blood clots and strokes and developmental delay.
11. Argininosuccinic Acidemia	An inherited disorder that causes ammonia to accumulate in the blood. High ammonia causes brain damage and can lead to coma. Treatment with a special diet can improve health outcomes.
12. Isovaleric Acidemia	A condition in the processing of certain amino acids. Acute illness and chronic disability can be prevented with a special diet.
13. Maple Syrup Urine Disease (including second-tier testing)	A condition in the processing of certain branch chain amino acids leads to a toxic build-up in the blood. If left untreated, infants can suffer severe neurological damage. Dietary treatment improves health outcomes.
14. Propionic Acidemia (including second-tier testing)	This group of amino acid disorders can present with acute episodes of illness during infancy and may cause progressive brain disease. Early detection and treatment offers the best chance of survival. However, developmental delay and other motor problems may occur despite treatment.
15. Methylmalonic Acidemia (including second-tier testing)	
16. Cobalamin disorders (CblA,B) (including second-tier testing)	

17. Cystic Fibrosis	A life-limiting disease that affects mainly the exocrine (mucus) glands of the lungs, liver and pancreas. Early detection and treatment of cystic fibrosis reduces the risk of mortality, prevents malnutrition, improves growth and may slow the progression of lung disease.
18. Sickle Cell Disease	A blood disorder characterized by red blood cells that assume an abnormal, rigid, sickle shape resulting in their restricted movement through blood vessels, depriving tissues of oxygen. Affected individuals experience lifelong periodic painful attacks and are at a risk of serious infections. Careful treatment and monitoring can improve health outcomes.
19. Congenital Adrenal Hyperplasia (to be added in October 2010)	A condition of deficient production of adrenal gland hormones. Early detection and treatment can prevent life-threatening dehydration.

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