



## Newborn Screening and PKU

### Backgrounder

#### **About Newborn Screening**<sup>i</sup>

- Newborn screening is a simple procedure conducted through bloodspot collection 24 to 48 hours after birth
- A newborn baby's heel is pricked in order to collect drops of blood for testing
- Blood samples collected are sent to a specialized laboratory to determine if a baby is likely to develop certain rare but treatable disorders, which can be life- or brain-threatening for the baby
- If the screening test is positive, further diagnostic tests will be conducted
- Once a condition is diagnosed, follow-up and treatment will begin immediately
- In some provinces, blood samples are stored after testing is complete for quality control, research and retesting purposes<sup>ii</sup>

#### **Newborn Screening in Canada**

- Newborn screening is recognized internationally as an essential, successful preventive public health program for early identification of disorders in newborns that can affect their long term health<sup>iii</sup>
- In Canada, mass newborn screening was first introduced in P.E.I. in 1963, in B.C. in 1964 and in Quebec in 1969<sup>iv</sup>
- In Ontario, Stephen Lewis, who was a Member of Provincial Parliament and Leader of the NDP at the time, was an advocate for a newborn screening program, and introduced a Private Member's Bill for newborn screening in the province
- Universal newborn screening began in Ontario in 1965, and in 2009, the program tested 144,947 babies<sup>v</sup>
- In B.C., about 40,000 babies are screened each year<sup>vi</sup>
- Today, each province and territory in Canada has their own government-run newborn screening program, screening for between 11 and 38 conditions – including Phenylketonuria (PKU)<sup>vii</sup>

#### **Newborn Screening and Phenylketonuria (PKU)**

- PKU was the first condition widely tested for through newborn screening
- If not detected, patients with PKU who go untreated are severely retarded, and may show challenging behavioral problems<sup>viii</sup>
- PKU and Congenital Hypothyroidism (CH) are the only two conditions universally screened for in every province and territory in Canada<sup>vii</sup>
- PKU screening has been embraced by industrialized countries, and is virtually universal in the United States, Western Europe, Australia, New Zealand, Israel and Japan<sup>ix</sup>

## **Guidelines for Newborn Screening and Treatment**<sup>x</sup>

- Newborn screening to detect treatable metabolic and other disorders is now an accepted standard of neonatal health care in almost all countries with well-developed medical services
- The International Society for Neonatal Screening (ISNS) is a non-profit organization that promotes the carrying out of appropriate newborn screening worldwide by sharing expertise and harmonizing programs, methods and protocols
- According to ISNS, newborn screening is recommended, provided that there is a satisfactory system in operation to deal with education, diagnostic testing, counseling, treatment and follow-up of patients identified by the test
- Shortly after screening programs were introduced in Canada, the World Health Organization stated that an important criterion for newborn screening tests is to provide appropriate treatment for any condition diagnosed<sup>xi</sup>
- Most newborn screening programs mandate that funding be made available for continuing treatment, if it is effective in modifying long-term adverse outcomes<sup>ix</sup>
- CanPKU believes that governments not only have a responsibility to screen for early identification of PKU, but also to uphold an important criterion of a screening test - to treat the condition adequately to improve health outcomes during the entire life of each person affected

### References

<sup>i</sup> Newborn Bloodspot FAQ's. Save Babies Through Screening Foundation. Available at: <http://www.savebabies.org/questions.html> Accessed March 9, 2012.

<sup>ii</sup> My Baby's Blood Sample. Newborn Screening. Available at: [http://www.newbornscreening.on.ca/bins/content\\_page.asp?cid=6-16-348&lang=1](http://www.newbornscreening.on.ca/bins/content_page.asp?cid=6-16-348&lang=1) Accessed March 28, 2012.

<sup>iii</sup> Overview: Newborn Screening. National Newborn Screening & Genetics Resource Centre. Available at: <http://genes-r-us.uthscsa.edu/resources/newborn/overview.htm> Accessed March 20, 2012.

<sup>iv</sup> Therrell BL, Adams J. Status of Newborn Screening in North America, *Journal of Inherited Metabolic Diseases*. 2007;30:447-465.

<sup>v</sup> 2009 Statistics. Newborn Screening Ontario. Available at: [http://www.newbornscreening.on.ca/bins/content\\_page.asp?cid=8-334-337&lang=1](http://www.newbornscreening.on.ca/bins/content_page.asp?cid=8-334-337&lang=1) Accessed March 28, 2012.

<sup>vi</sup> BC Newborn Screening Program. BC Women's Hospital & Health Centre. Available at: <http://www.bcwomens.ca/Services/PregnancyBirthNewborns/NewbornCare/NewbornScreeningProgram/default.htm> Accessed April 3, 2012.

<sup>vii</sup> Newborn Screening in Canada Status Report. Canadian PKU and Allied Disorders Inc. Available at: [http://raredisorders.ca/documents/CanadaNBSstatusupdatedApril182012\\_000.pdf](http://raredisorders.ca/documents/CanadaNBSstatusupdatedApril182012_000.pdf) Accessed April 20, 2012.

<sup>viii</sup> Trefz F, Maillot F, Motzfeldt K, Schwarz M. Adult phenylketonuria outcome and management, *Mol. Genet. Metab*. 2011. doi:10.1016/j.jymgme.2011.08.025.

<sup>ix</sup> Hanley, William B. Newborn screening in Canada – Are we out of step? *Paediatr Child Health*. April 2005; 10:4:203-7. Available at: <http://www.ncbi.nlm.nih.gov/pmc/articles/PMC2722527/> Accessed March 20, 2012.

<sup>x</sup> ISNS General Guidelines for Neonatal Screening. International Society for Neonatal Screening. Available at: <http://www.isns-neoscreening.org/htm/generalguidelines.htm> Accessed March 9, 2012.

<sup>xi</sup> Wilson JMG, Jungner G. Principles and Practice of Screening for Disease. Geneva, Switzerland: World Health Organization; 1968:27. Available at: [whqlibdoc.who.int/php/WHO\\_PHP\\_34.pdf](http://whqlibdoc.who.int/php/WHO_PHP_34.pdf) Accessed March 20, 2012.