Newborn Screening and PKU

Backgrounder

About Newborn Screening

- 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

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
- In Canada, mass newborn screening was first introduced in P.E.I. in 1963, in B.C. in 1964 and in Quebec in 1969
- 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
- In B.C., about 40,000 babies are screened each year
- 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)

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
- PKU and Congenital Hypothyroidism (CH) are the only two conditions universally screened for in every province and territory in Canada
- PKU screening has been embraced by industrialized countries, and is virtually universal in the United States, Western Europe, Australia, New Zealand, Israel and Japan
Guidelines for Newborn Screening and Treatment

- 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.
- Most newborn screening programs mandate that funding be made available for continuing treatment, if it is effective in modifying long-term adverse outcomes.
- 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