Second Opinion

Falling behind in screening babies

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Within hours of birth, every newborn gets the infamous heel prick -- a few drops of blood drawn and smeared onto a special piece of paper. Those samples are sent to a laboratory, where they are tested for rare genetic disorders.

The number of tests performed varies wildly between jurisdictions, from a low of two in Ontario to a high of 28 in Saskatchewan. (In the United States, the range is more pronounced, from six in West Virginia to 75 in California.) There is no ideal number of routine screening tests that should be performed on newborns.

The best evidence currently available comes from an April report by the American College of Medical Genetics. That advisory group recommended that 29 tests be performed routinely on all babies -- a hearing test and 28 blood tests for conditions such as sickle cell anemia and PKU (phenylketonuria, which is an inability to process the amino acid phenylalanine that then accumulates and damages the brain).

In Canada, only Saskatchewan performs all the recommended tests. Ontario has the dubious distinction of conducting fewer screening tests than any other jurisdiction in the developed world. When newborn screening started 40 years ago, Canada was a world leader.

Now it has a dismal record, lagging behind every developed country, and many in the developing world as well.

In many ways, this is a classic dilemma of public health. Should we spend money testing for rare conditions when health dollars may have a greater impact in other areas, like ensuring that all pregnant women get adequate folic acid?

While it is true that many of these conditions are rare, they are also silent stalkers. Screening tests are necessary because the damage -- more often than not severe mental retardation -- can begin and end before the first clinical signs of illness appear. Then, it's too late.

The ACMG report was careful to recommend testing only for conditions that meet three criteria: There is a reliable, affordable test; there is a clear understanding of the effects of the condition; there are interventions (diets and drugs, for example) that can benefit a child who is diagnosed.

Failing to act under those circumstances seems unconscionable.

A child with undiagnosed PKU will be severely mentally retarded by age 1. With testing and treatment (a nutritional supplement and a low-protein diet), that same child will grow up normal.

A baby with congenital hypothyroidism will suffer stunted growth and grave developmental problems, but treatment with a single pill daily will avoid those symptoms.

At least Ontario screens for those two disorders, as well as hearing. But why not the others?

If a baby dies under the age of 2, the tests are conducted routinely to determine the cause of death. (It is believed that some cases of sudden infant death syndrome, SIDS, are actually undiagnosed metabolic disorders.) But shouldn't Ontario be testing its live babies, not just the dead ones?

It is incomprehensible, too, that a province whose capital, Toronto, is one of the most multicultural cities in the world, does not screen every newborn for sickle cell anemia and thalassemia, two common blood disorders (notably among blacks and those of Mediterranean descent, respectively), which can damage vital organs and leave children prone to life-threatening infections. (So far, only the Scarborough Hospital does such testing routinely.)

It is also difficult to understand why Ontario and other provinces have not embraced the technology, if for no other reason than it is cost-effective.

The Save Babies Through Screening Foundation estimates that if Ontario did the recommended 29 tests, it would cost about $2-million a year.

Among the 130,000 births in the province, about 160 additional cases of genetic disorders would be identified, and those children would be spared the devastating effects of these conditions (or, at the very least, have their symptoms mitigated).

While many of the rare conditions are expensive to treat, U.S. research suggests late detection (waiting until symptoms appear) results in treatment costs that are about four times as high.

Consider that the medical costs of a child with severe mental retardation exceed $1-million, not to mention the costs to the education system and the impact on the family.

We cannot lose sight either of the fact that this is just a dress rehearsal. Making decisions about 29 screening tests is easy. The technology exists today to do thousands of genetic tests. The time to develop a decision-making process, a sound way of evaluating the cost-effectiveness and ethical rules for newborn screening is now.

Rapid changes in technology mean new standards of care are required to keep pace. Children in Canada should not be left behind. Babies, regardless of where they are born, should have equal access to screening and services.

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