Pacific Northwest Regional Newborn Screening: A Paradigm of Prevention*

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We describe the use and early results of the Oregon Public Health Laboratory computer to monitor hospital and practitioner compliance with state newborn screening recommendations. The system tracks five major categories of screening practice in which 18 types of errors affecting screening test quality have been identified. Results show that 47.3% of newborn screening specimens were submitted incorrectly according to current state recommendations. The program will be used to improve screening practices and to evaluate educational efforts.

INTRODUCTION

In 1963, Guthrie and Susi described "A Simple Method for Detecting Phenylketonuria in Large Populations of Newborn Infants."1 In the last 30 years, newborn screening has evolved from these "simple" beginnings into the largest, most complex public health program ever undertaken by civilized countries. Today over 50 different disorders can be detected in dried blood spots using the bacterial inhibition assay or other methods, such as fluorometry or radioimmune assay. In the United States, 37 separate programs screen 3.5 million infants a year for congenital hypothyroidism and phenylketonuria. Many programs also screen for additional disorders such as galactosemia or sickle cell disease.

Approximately 1000 infants a year will be born with one of the disorders covered by screening programs. The vast majority of these infants will be correctly identified by screening tests and referred for diagnosis and treatment, saving millions of dollars and...
untold misery for the children and their families. The small number of infants missed by screening and the enormous liability incurred for their life-time of care has focused increasing attention on quality control issues.

Newborn screening is a process which requires coordinated effort from three groups of health care providers:

1. **Practitioners:** responsible for collection and handling of screening specimens, parent education and prompt action on any abnormal result notified to them. We refer to the practitioner’s participation in the program as NEWBORN SCREENING PRACTICE.

2. **Central Screening Laboratory:** responsible for testing, record keeping, quality control of laboratory methods, notification of results and tracking of abnormal and unresolved results.

3. **Treatment and Follow-up Team:** responsible for the confirmation of infants with abnormal results, management of confirmed cases and for education of practitioners.

Most state and regional programs have devised quality assurance measures for the laboratory and follow-up components of their screening programs. These include centralized laboratories who employ advanced analytical methods, designated follow-up coordinators, and the use of computers which link laboratory and follow-up teams into a unified force.

In contrast, no adequate method of quality assurance has yet been devised for newborn screening practice, increasing the chance that affected infants will escape detection. A recent report indicated that 1:70 cases of phenylketonuria and 1:120 cases of hypothyroidism had been missed by screening programs in the United States; one third of these were due to problems in screening practice.² The need to monitor screening practices has become particularly acute because of two major changes in perinatal care—one increase in out-of-hospital births and the early discharge of infants from hospitals. In Oregon, for instance, 20% of infants are discharged within 24 hr of birth and 70% within 48 hr of birth. According to Oregon State Health Division records from birth certificates, 5% of infants are born in nonhospital birthing centers or at home.

We have developed a computer-based surveillance system which tracks: individual infants through the screening process to ensure timely and complete follow-up; screening practices both in hospitals, birth centers, and by individual practitioners so that those with high error rates can be identified; and changes in screening practice error rates over time so that educational interventions can be evaluated.

This paper outlines the computer surveillance of 18 screening practice errors in Oregon, how these data will be used to improve screening practices, and some of the steps required for the development of the program.

The Oregon State Public Health Laboratory provides newborn screening for all infants born in Oregon, Idaho, Nevada, and Alaska. The Pacific Northwest Regional Newborn Screening Program encompasses the largest geographic area in the country, with close to 1 million square miles. Each year, approximately 135,000 specimens are submitted from 85,000 infants. The screening battery includes assays for seven disorders: congenital hypothyroidism, hyperphenylalaninemia, galactosemia, maple syrup urine disease, homocystinuria, tyrosinemia, and biotinidase deficiency. In addition, Alaskan specimens are assayed for congenital adrenal hyperplasia. Tests for sickle cell and cystic fibrosis are under consideration.