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Expanded metabolic screening utilizing tandem mass spectrometry: The Massachusetts experience in the first year.

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From February through November 1999, the first 60,000 newborns (of an annual birth cohort of 80,000) in MA were screened by tandem mass spectrometry (MS) for 20 metabolic disorders, as an expansion beyond 3 amino acids (phenylalanine, methionine, leucine) and 6 other routine screens: galactosemia, biotinidase deficiency, hypothyroidism, CAH, hemoglobinopathy and toxoplasmosis. [Screening methods for the latter 6 were unchanged.]

The MS expanded screening disease categories included:

a. Amino Acids (AA): arginine, ornithine, citrulline (urea cycle defects); tyrosine (tyrosinemia type 1 and 2). No cases were identified.
b. Organic Acids (OA): propionic (PPA), methylmalonic (MMA) and glutaric acidurias (GA1 or 2), HMG Co A lyase deficiency, ketothiolase deficiency. Two PPA were identified.
c. Fatty Acid Oxidation (FOD) defects: short, medium and long chain acyl-Co A dehydrogenase deficiencies (SCAD, MCAD, LCAD, LCHAD, VLCAD) and carnitine palmitoyl transferase deficiency (CPT-2). Two SCAD, one MCAD and one CPT-2 were identified.

Of the 6 detections, 5 were asymptomatic. The infant with CPT-2 deficiency died the day the blood spot was received.

Conclusion: By MS, the 3 original AA were detected in the usual frequency. For OA, the prevalence was approximately as expected, MCAD was less than expected, and SCAD may be more common than we had inferred from rare published reports.

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Newborn screening for sickle cell disease: assessing program effectiveness.

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Children affected with sickle cell disease (SCD) are at increased risk for morbidity and mortality, especially in the first three years of life. Newborn screening programs for SCD currently operate in over 40 states. To assess the effectiveness of newborn screening in providing early medical interventions and in reducing morbidity and mortality, a three-year follow-up study was conducted in 1998 in three states (California, Illinois, and New York), where universal newborn screening for SCD is offered.

1042 children born in 1992 and 1993 were diagnosed with SCD in California (n=265), Illinois (n=254), and New York (n=523). 61% (n=634) of children were diagnosed with SS disease, and 32% with SC disease (n=328). Of children born and diagnosed with SCD in 1992 and 1993, fourteen were deceased at the time of the study. A parental survey and provider survey were administered for each child to examine genetic factors, medical care and compliance, and sociodemographic factors. Provider surveys were completed for 72% (n=752) of the children and parental surveys were completed for 24% (n=252) of the children. Data from both surveys were merged; both parental and provider surveys were completed for 18% (n=184) of the children. Preliminary results indicate that 44% of respondents were informed of SCD services available for their child with 27% having utilized these services. Penicillin was taken regularly by 93% of respondents; 75% had received pneumonia vaccine or pneumovax, and 65% had received a full series of HIB/tetramune. Finally, 44% of SCD children reported compliance with prophylactic antibiotic regimen. Associations of these and other factors with various morbidity measures are currently being assessed in the entire population as well as by state.

This study utilized two methods for ascertaining follow-up and made repeated attempts to locate the children. Poor response rates for both provider and parental surveys demonstrate the need for on-going systematic collection of follow-up data on children with sickle cell disease to identify the extent and gaps of delivering proper medical services and interventions.