

Recent evolution of newborn screening – A view from a state advisory committee

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The early to mid 1990's

- Ensuring access to medical foods
- Refining methods for already existing tests
- Developing parent information brochures
- Refining general criteria for adding new tests
- Ongoing issues with unsatisfactory samples
- Dealing with MCOs about follow-up testing
- Ensuring appropriate emergency follow-up

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The MCAD story – the early days

- 1992 – mutation study using left over blood spots showed carrier frequency of 1/249 – deemed insufficient to support newborn screening
- 1993 – autopsy study conducted in SIDS cases to look for MCAD
- GAC decided data at that time were not sufficient to proceed

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The MCAD story – the next phase

- 1998 -- Letter from parents of child with MCAD sent to prominent state senator urging adoption of MS/MS on behalf of FOD (Fatty Oxidation Disorders Support Group)
- GAC decided against adopting MS/MS at that time, in large part because “[s]everal of the diseases detected have no known treatment” and therefore did not meet all the screening criteria
 - Evidence for CAH (2000) and biotinidase deficiency (2003) more compelling

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The MCAD story

- Institution of the Massachusetts pilot study
- 2000 – TDH and GAC memo -- monitor other pilots, expressed concerns about
 - Rarity and lack of rx for many disorders identified by MS/MS,
 - Cost-effectiveness, and
 - Lack of funds for follow up
- 2000 – GAC recommended adding MCAD, MSUD, and homocystinuria, responding in part to March of Dimes

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The MCAD Story

- 2001 – MMWR report noting impact of parent advocacy groups despite limited data in medical literature
- 2001 – TN decides to move toward adopting MS/MS at least for MCAD
- 2002 – Bill introduced in Mississippi requiring screening for all disorders detectable by MS/MS, driven by advocacy by Vince and Robin Haygood
 - TN no longer performs newborn screening for Mississippi

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TN SB0056 (2003)

(c) Regulations promulgated by the department pursuant to this section shall not preclude the use of private laboratories, as an alternative to state government operated laboratories, to conduct the analysis required by subdivision (a)(1) so long as such laboratory meets the following requirements.

(1) The laboratory must be certified according to the federal Clinical Laboratory Improvement Amendments of 1998, 42 U.S.C. Section 263(a), and must be licensed to perform screening testing of newborn infants in the state in which the laboratory is located and such license must be in good standing.

(2) The laboratory must provide screening for at least the following:

- (A) Phenylketonuria;
- (B) Galactosemia;
- (C) Hypothyroidism;
- (D) Sickle Cell Anemia and other hemoglobinopathies;
- (E) Cystic Fibrosis;
- (F) Congenital Adrenal Hyperplasia;
- (G) Maple Syrup Urine Disease;
- (H) Homocystinuria;
- (I) Biotinidase Deficiency; and
- (J) Medium Chain Acyl-Coa Dehydrogenase Deficiency.

(3) The laboratory must agree to provide the division of maternal and child health with prompt reports and data to ensure that the patient has confirmatory testing, diagnosis and treatment.

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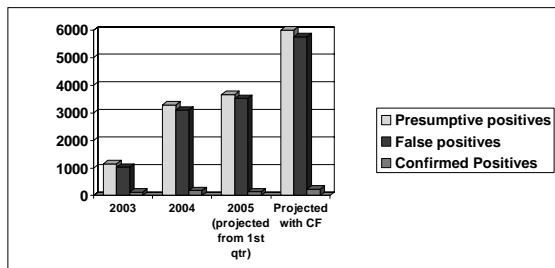
The response

- Talk with legislators
- May 2003 – decide to proceed with reporting all analytes detectable by MS/MS
- October 2003 – machines in place
- January 2004 – screening begins, initially for MCAD, MSUD, and homocystinuria
- Summer 2004 -- analytes for 50 disorders are reported
- Spring 2005 – decided to proceed with CF
Debate about IRT/IRT v. IRT/DNA

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What is the impact of expanded screening in TN?



~80,000 births per year

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What are the issues?

- What are the goals of screening?
 - The new disorders do not fit the old paradigm
 - Much more emphasis on parental desire and perception that knowledge is good per se
- Is there any way to limit newborn screening?
 - State run
 - Private
- Should we care?



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