

Expanding Newborn Screening: The Minnesota Model



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Organization and Guidance

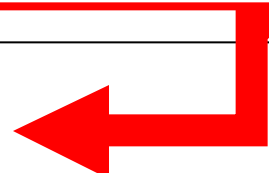
- **MN Statutes 2003, Chapter 144.125-128 and
MN Rule 4615.0300 – 4615.0760**
- **Mandated newborn screening advisory
committee**
- **Health Commissioner oversight**
- **Guidance from WHO, AAP, APHL, NNSGRC
and ACMG**



Minnesota Timeline

Year	Disorder	Incidence
1965	Phenylketonuria	1:13,500 (~5 per year)
1967	Congenital Hypothyroidism	1:2,500 (~23 per year)
1974	Galactosemia	1:60,000 (~1 per year)
1988	Sickle Cell Anemia (+ others)	1:3,800 (~17 per year)
1993	Congenital Adrenal Hyperplasia	1:17,000 (~4 per year)

>1,300 newborns diagnosed in MN





2001: A Year of Changes

Expansion begins

Congenital Hearing Loss

- Voluntary screening
- 108 / 111 birthing centers
- 98% infants screened

Tandem Mass Spectrometry (MS/MS)

- 204,000 infants screened
- 90 confirmed positives
- Aggregate incidence ~ 1:2,300



Clinical Impact of Expanded Screening Using MS/MS

Year	2001	2002	2003
Screened	39,974*	69,821	71,147
Abnormal	247	277	172
True Positives	11	28	46
False Positives	236	249	126

* 7 months

1:3,634

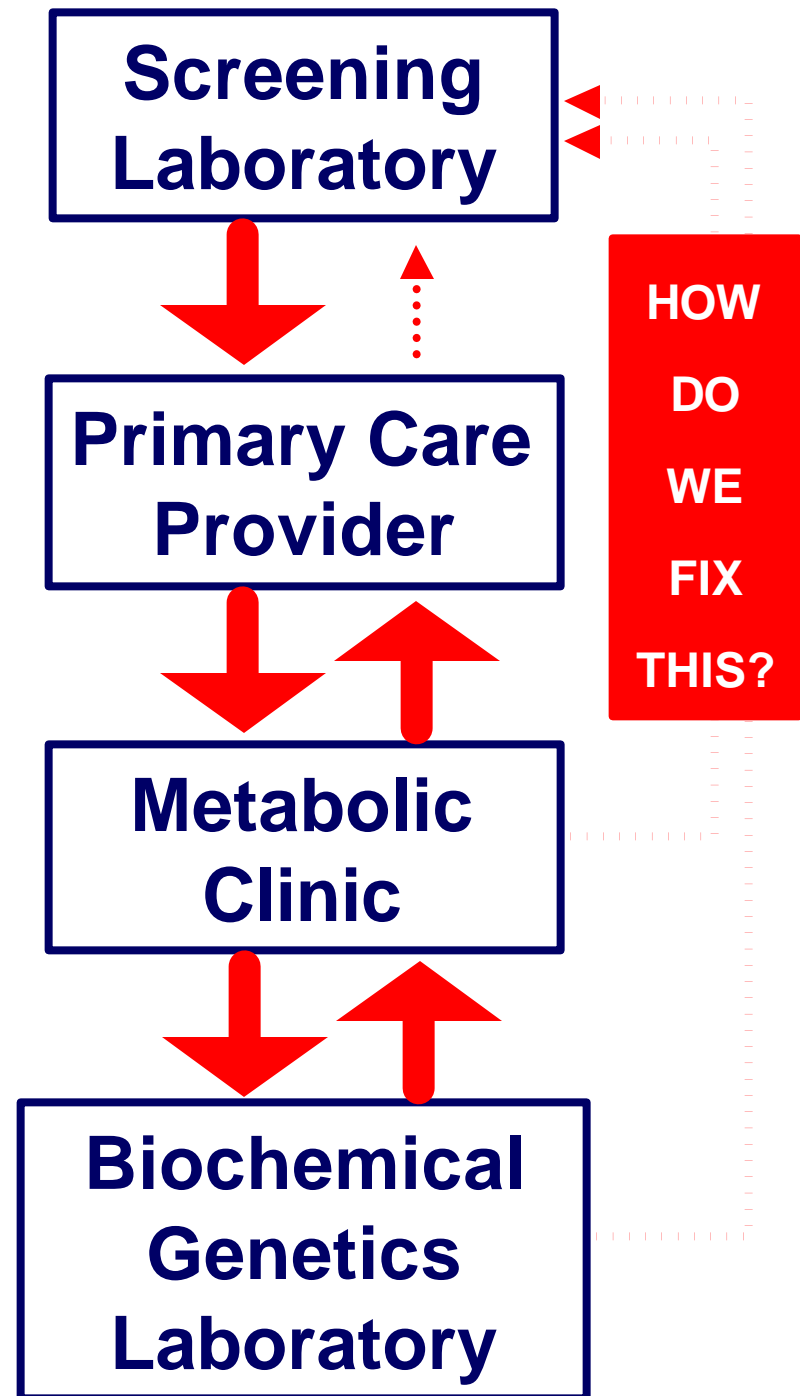
1:2,493

1:1,547



Challenges

- Minimal funding
- Sustainability of expanded screening
- Limited outcome tracking and follow-up
- Limited support for primary care, infant and family
- **LACK OF COMMUNICATION**



The Minnesota Model



What is the Minnesota Model?

A multi-component newborn screening program based on **COORDINATION** and **COMMUNICATION** between all parties involved with the goal to expand and enhance the delivery of patient care, laboratory tests, and family services

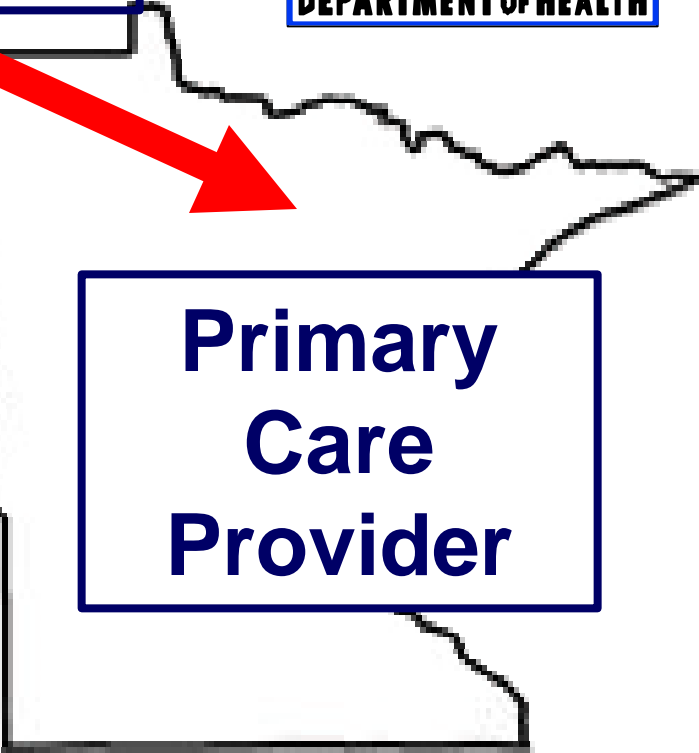
Coordination & Communication (The Minnesota Model)

- **Notification of abnormal results**
- **Verification**
- **Reporting**
- **Referral of true positives**
- **Follow-up**

Notification

- Phone
- Fax
- E-mail

**Screening
Laboratory**



**Biochemical
Genetics
Laboratory**

**Primary
Care
Provider**

**Metabolic
Clinic**



Verification of Abnormal Results

Screening Laboratory

Discourage repeat submission and testing of blood spots

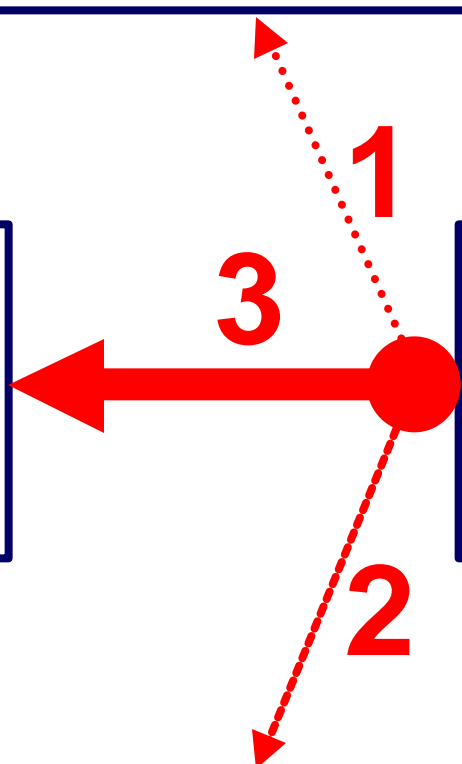
Biochemical Genetics Laboratory

Primary Care Provider

Activate rapid confirmatory testing

Metabolic Clinic

Prevent "blind" referrals to specialists of false positives



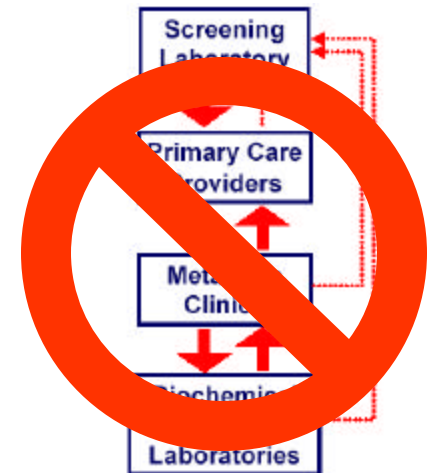
Reporting of Diagnostic Testing

**Screening
Laboratory**

**Biochemical
Genetics
Laboratory**

**Primary
Care
Provider**

**Metabolic
Clinic**



**Referral
of TRUE
Positives**

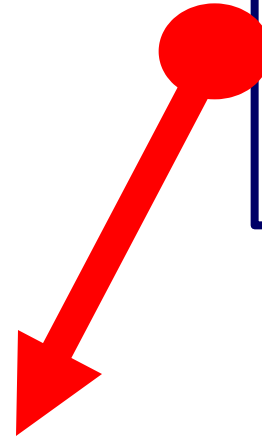
**Screening
Laboratory**

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**Primary
Care
Provider**

**Metabolic
Clinic**

**Confirmatory
testing is
already DONE**



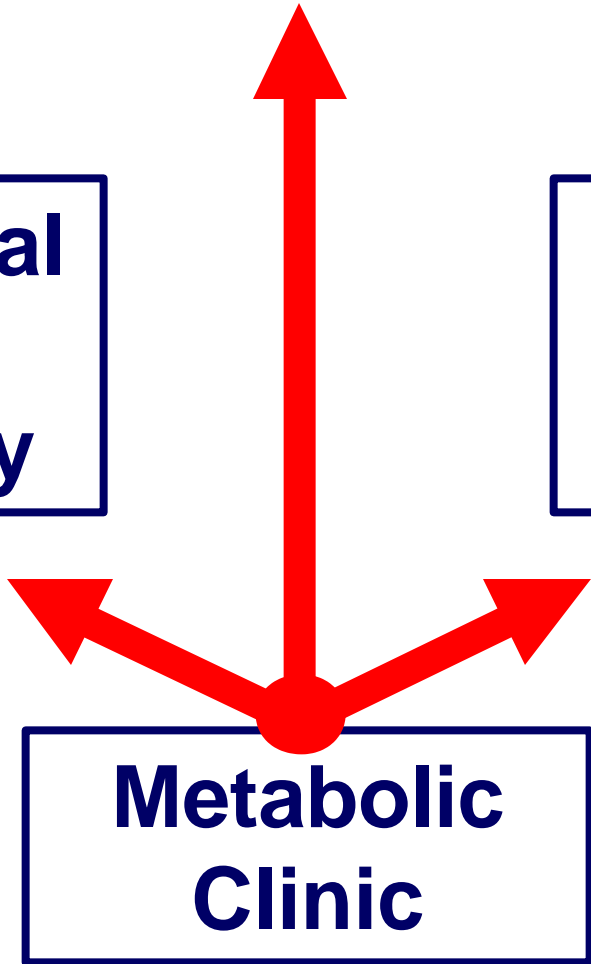
SHORT Term Follow-Up

**Screening
Laboratory**

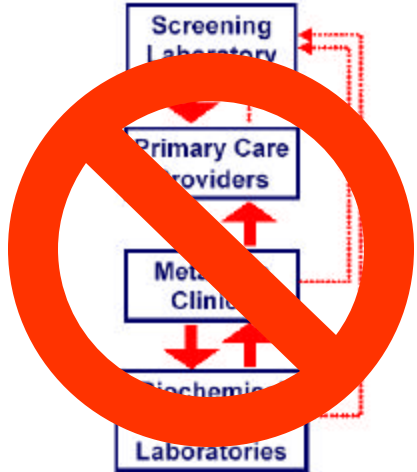
- BENEFITS
- Accurate statistics
 - Monitoring of trends
 - Learning from experience
 - Education

**Biochemical
Genetics
Laboratory**

**Primary
Care
Provider**



**Metabolic
Clinic**



Joint Newborn Screening Program in Minnesota (May 2004)

Established Conditions



Cong. Hypothyroidism	S,FU	-
Galactosemia (GALT)	S,FU	-
Sickle Cell Anemia	S,FU	-
21-Hydroxylase Def (CAH)	S,FU	2T
MS/MS (AA, OA, FAO)	FU	S,C

S Screening
FU Follow-up

C Confirmatory
2T 2nd tier test in DBS

Joint Newborn Screening Program in Minnesota (2004-2005)

Future Additions



Biotinidase Deficiency

D,S,FU

C

G6PD Deficiency

D,S,FU

-

Wilson Disease

S,FU

D,P

Others

?

?

D Development

S Screening

C Confirmatory

P Pilot Study

FU Follow-up

Acknowledgements



Chet Whitley

Amy Hietala

Dietrich Matern

Lisa Schimmenti

Chris Brueske

Si Houn Hahn

Dorothy Markowitz

Jill Simonetti

April Studinski

Kristi Bentler

Dan Johnson

Kara Mensink

Cindy Lorentz

Carolyn Anderson

Jean Kroning

Mary Ahrens

Dana Brown

Angela Tauscher