

# Utilization of Follow-up Services Following MS/MS Screening Referral

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# Introduction

- Metabolic centers provide follow-up services to infants with positive test results under contract to the Genetic Disease Branch, CA Department of Health Services.
- As a result of the 18 month MS/MS Pilot Screening Program a new group of newborns were referred.
- The newborns required specialty metabolic services to determine their disease status.

\* All data is preliminary pending further analysis

# Problem

We need to know more about the nature, scope and effectiveness of follow-up services following the implementation of a new MS/MS screening program.

\* All data is preliminary pending further analysis

# Objectives

- To evaluate the nature and scope of the metabolic services provided to California's newborns following the implementation of the MS/MS pilot screening program.
- To develop a data collection tool to collect clinical follow-up services data. (One form for every patient contact.)
- To study several aspects of the breadth and depth of the services that were provided (services provided, tests ordered, baby health status: at first and subsequent visits to metabolic centers)

\* All data is preliminary pending further analysis

# Evaluation Goals

- Document the type of tests and services provided
- Examine the effectiveness and efficiency of the follow-up process
- Inform the development of follow-up services guidelines
- Use data to estimate the costs of MS/MS follow-up services.

\* All data is preliminary pending further analysis

# Method

## The Patient Contact Form<sup>(1)</sup>

### Data collected included:

- Who provided follow-up services (physician, nurse, genetic counselor, etc)
- Mode of contact (phone vs. in person)
- Type of appointment (routine follow-up visit, education session, etc)
- Length of each contact (in minutes)

\* All data is preliminary pending further analysis

# Method

## The Patient Contact Form (2)

### Data collected included:

- Diagnostic laboratory tests ordered
- Whether new treatments initiated or changed
- Assessment of health status at the time of the patient contact
- Inventory of parental concerns

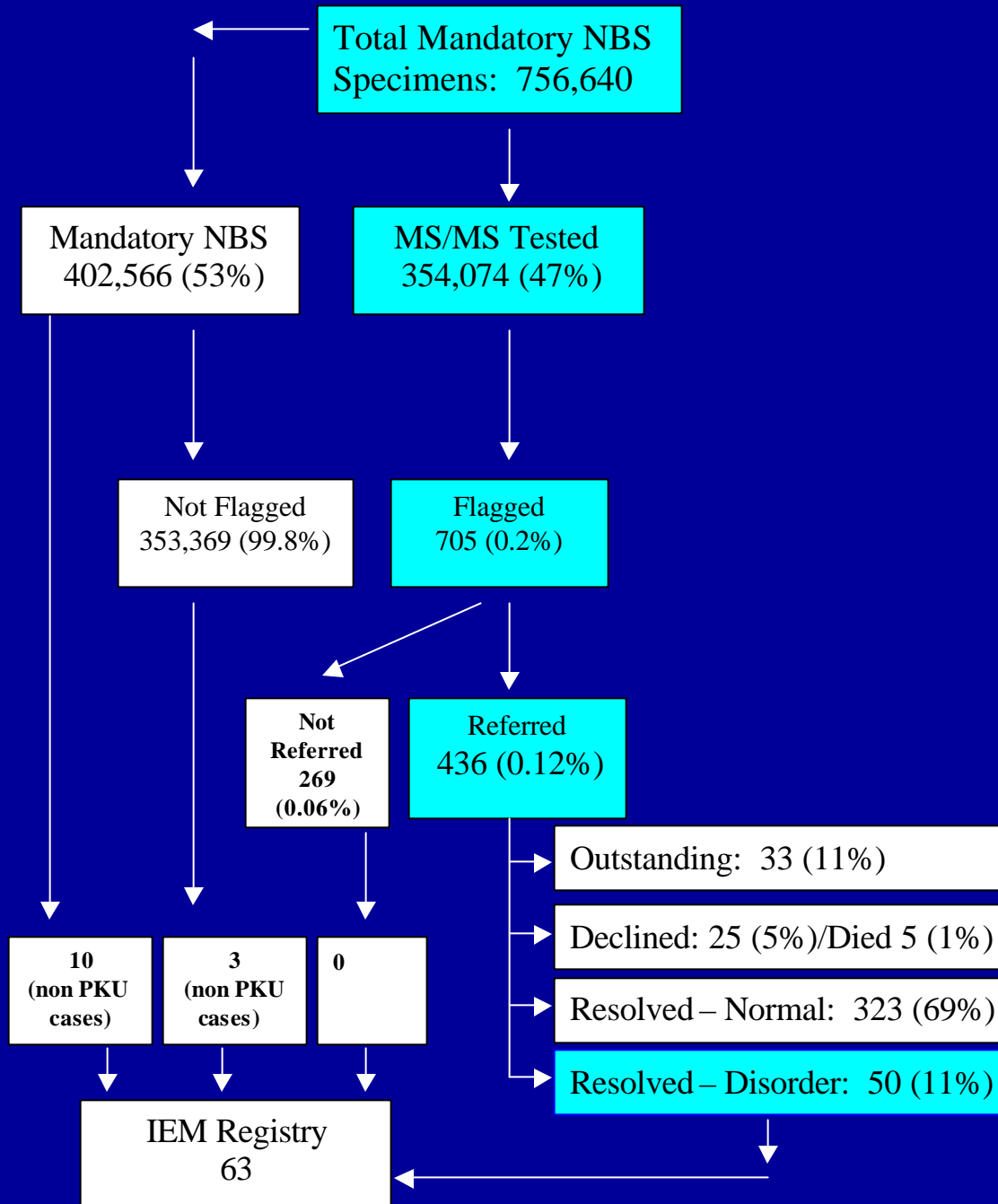
\* All data is preliminary pending further analysis

# Method

**Newborns were grouped by whether or not the baby had any health problems at the time of the first patient contact.**

- What tests were ordered?
- What were their results?
- Diagnostic status of newborn?

# Summary of MS/MS Screening



# Overall MS/MS Test Performance

- One newborn was referred for every 800 babies screened;
- One disorder was successfully diagnosed for every 7000 newborns screened
- One disorder was diagnosed for every 9 infants referred.

\* All data is preliminary pending further analysis

# Overall MS/MS Pilot Test Performance

**50 confirmed disorders were reported**

|         |                             |
|---------|-----------------------------|
| MCADD:  | 13 cases among 15 referrals |
| SCADD:  | 18 cases among 90 referrals |
| MMA/PA: | 11 cases among 51 referrals |
| 3MCC:   | 3 cases among 25 referral   |
| GA-1:   | 1 case among 5 referrals    |
| GA-II:  | 1 case among 2 referrals    |
| MSUD:   | 1 case among 40 referrals   |
| VLCADD: | 1 case among 2 referrals    |

\* All data is preliminary pending further analysis

# Suspected Diseases

## Among 436 referrals

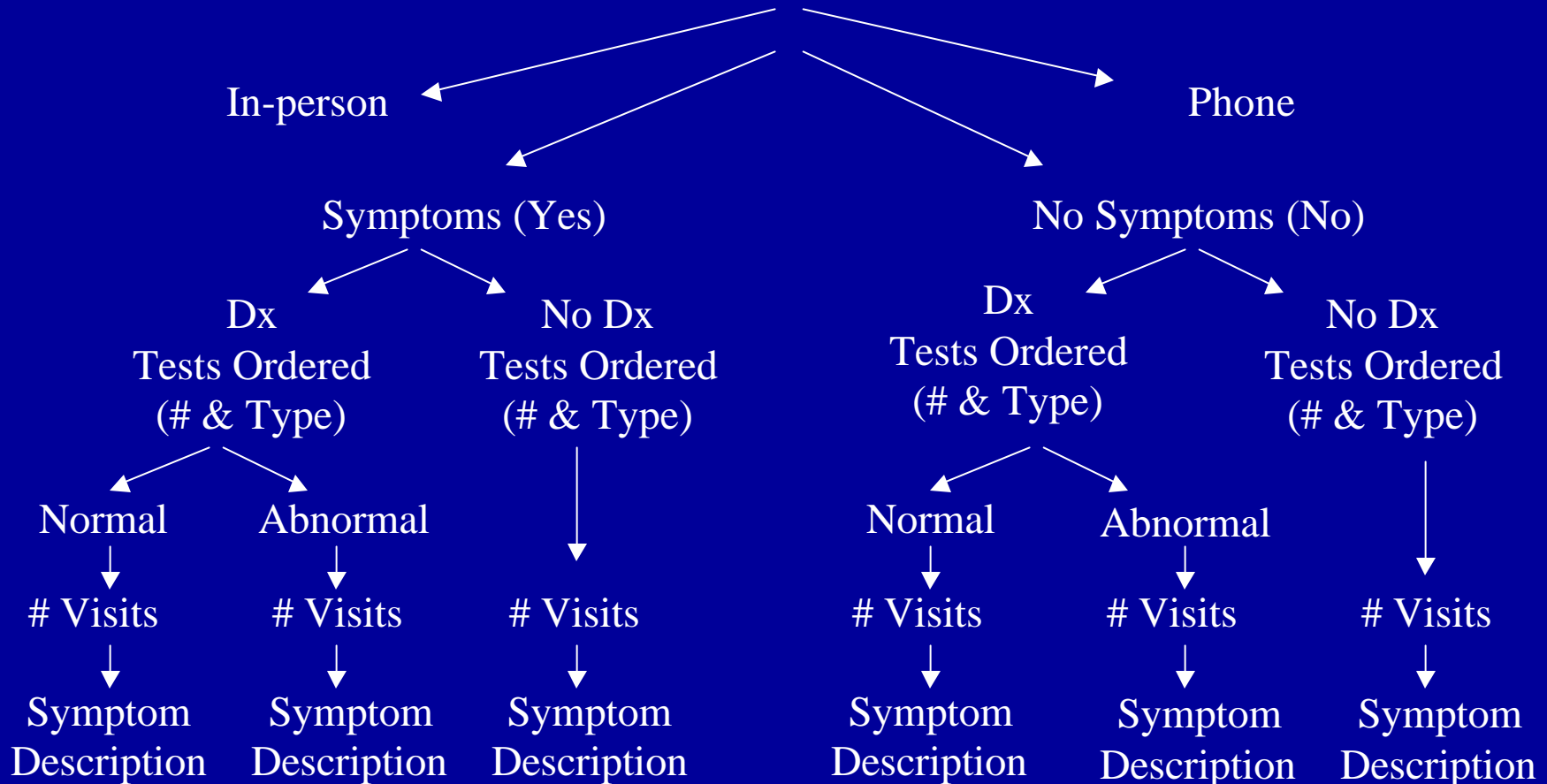
0.12% of all of MS/MS screened patients were referred for follow-up.

- 20% - Suspected SCADD
- 12% - Suspected MMA/PA
- 11% - Suspected Homocystinuria
- 9% - Suspected MSUDD
- 6% - Suspected 3MCC
- 5% - Suspected Argininemia
- 5% - Other Amino Acid Disorders
- 3% - Suspected MCADD
- 9% - *Non-specific disorder*

\* All data is preliminary pending further analysis

# Flow Chart Model

## MS/MS Referred Cases (First Visit/Contact)



\* All data is preliminary pending further analysis

# Types of Flow Charts

All Referrals Combined

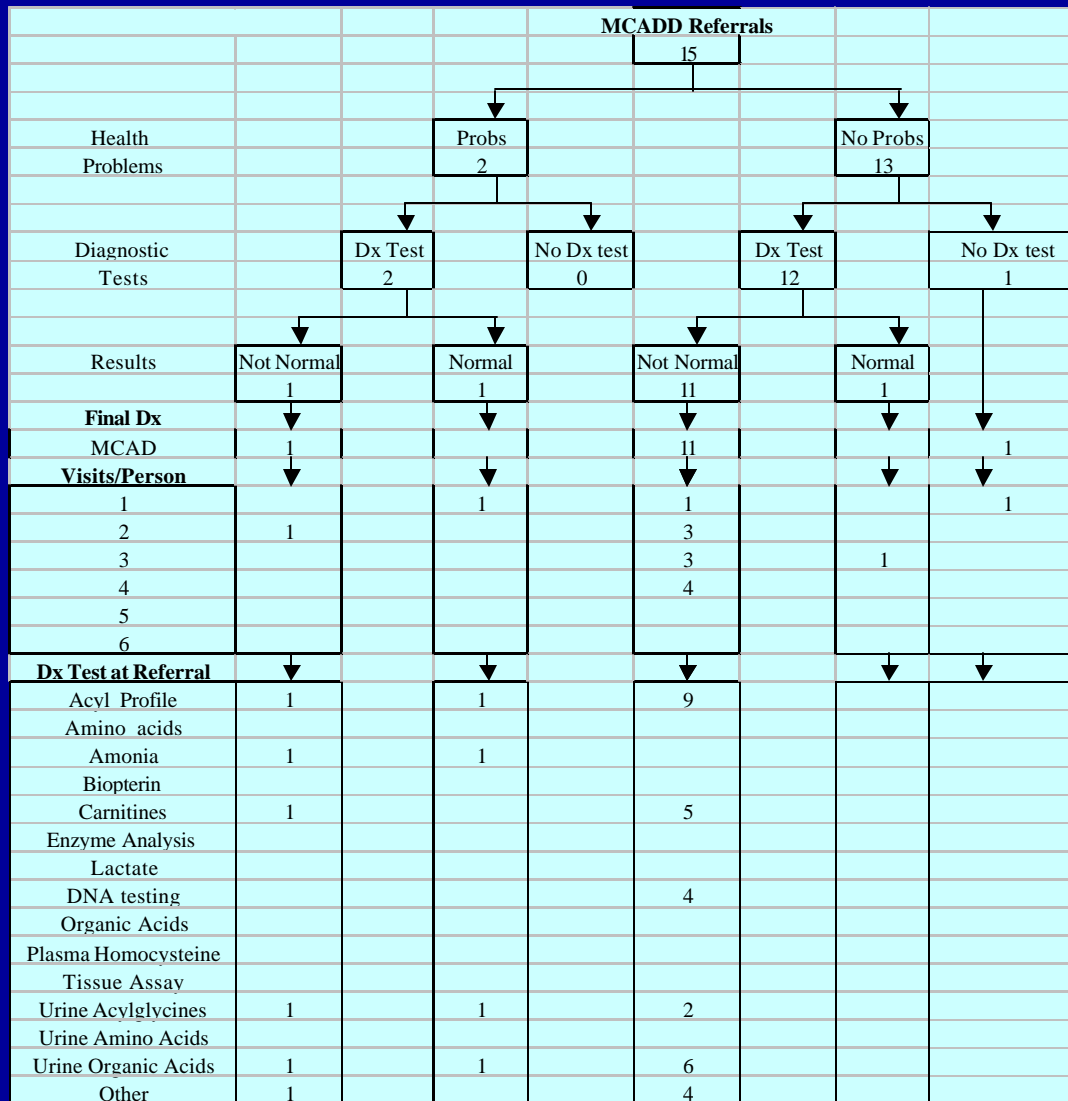
All Disorders Combined

False Positives Combined

Specific Disorders

\* All data is preliminary pending further analysis

# MCADD Referral Flow Chart



\* All data is preliminary pending further analysis

# The First Patient Contact

- A majority (58%) of the referrals were not seen at the metabolic center
- Centers were able to work with the newborns' primary care physicians to order and evaluate initial diagnostic tests.

\* All data is preliminary pending further analysis

# Patient Contact Summary

**436 patients referred: 609 Patient Contact Forms**

- 70% had one contact with the metabolic center  
(19% for MCADD)
- 16% had two contacts  
(25% for MCADD)
- 7% had three contacts  
(31% MCADD)
- 4% had four or more contacts  
(25% MCADD)

\* All data is preliminary pending further analysis

# Health Problems

(first visit)

**10% of all referrals were had health problems during the first program visit**

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- 13% had symptoms among suspected MCADD referrals
- 15% had symptoms among suspected SCADD referrals
- 17% had symptoms among suspected Other Disorders Group referrals

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# Diagnostic Tests Orders

(first visit)

- 91% had at least one type of diagnostic test ordered
- 751 laboratory tests were ordered for 436 patient referrals
- 21% of the referrals had an acylcarnitine profile test
- 28% had an amino acid profile
- 26% had a urine organic acid test ordered

\* All data is preliminary pending further analysis

# Abnormal Diagnostic Tests

(First Visit)

*Among patients with symptoms:*

50% had one or more abnormal diagnostic test result

*Among patients with no symptoms:*

32% had one or more abnormal diagnostic test result

\* All data is preliminary pending further analysis

# Conclusion

**Today's presentation reflects only some of the data collected as part of the MS/MS follow-up services utilization study.**

Additional issues to be explored include:

- \* Description of services in subsequent visits
- \* Summary of parental concerns
- \* Assessing the costs of follow-up services:
  - summary of type and length of visits
  - summary of providers delivering service
  - summary of treatments and monitoring tests

\* All data is preliminary pending further analysis

# For More Details on Project Findings

Visit our web-site at  
[www.CaliforniaMsMs.org](http://www.CaliforniaMsMs.org)

**Special thanks to staff at the Genetic Disease Branch and  
all California providers, patients and contract staff  
who made this experience possible.**

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