

California Newborn Screening Program

Long Term Follow-up Data System for Metabolic Disorders

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California

Long Term Follow-Up Data System

**Surveillance of diagnosed cases through age five
using an annual survey instrument:**

Metabolic Center Annual Patient Summary (MCAPS)

- Availability of ongoing care and management
- Clinical outcomes and developmental assessment
- Impact on health care utilization



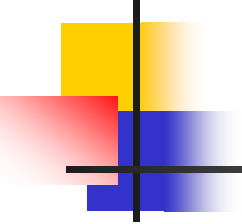
A Brief History

Summer, 2005:

- Web-based Screening Information System (SIS) go-live
- MS/MS screening go-live

Summer, 2007:

- Implementation of long term follow up data system using Metabolic Center Annual Patient Summaries (MCAPS)



The Metabolic Center Annual Patient Summary (MCAPS)

- Yearly assessment, or “snapshot,” to capture status of child at the end of each completed year of life
- Web-based Screening Information System (SIS) adds each child’s name to the “MCAPS pending case list” after the child’s birthday
- After the report is completed the name falls off the pending list and will reappear the following year - through age five.



MCAPS Data Elements

- **Clinic follow-up status**
 - **Services provided by the metabolic center in previous year**
 - **Date of last visit/interaction with patient that occurred in previous year**
 - **Total number of patient visits to the metabolic center in previous year**
 - **Total number of hospitalizations/emergency room visits, with length of stay and reason for admission, in previous year**
 - **Has patient been symptomatic in previous year?**
 - **Persistent health problems/symptoms in previous year**
 - **Treatments/therapies/strategies prescribed in previous year**
 - **Assessment of patient's development and function (speech/cognitive/physical/fine motor/gross motor)**
 - **Loss of skills that had been previously achieved**
 - **Global Health Assessment (scale: 1=critical through to 6=excellent)**

Count of MCAPS Entered in SIS by Disorder Type & Age of Child (8/1/2007 – 2/9/2009)

| | 1 st Year | 2 nd Year | 3 rd Year | 4 th Year | 5 th Year | All |
|-------------|----------------------|----------------------|----------------------|----------------------|----------------------|-----|
| 3MCC | 19 | 14 | 4 | . | . | 37 |
| ARG | . | 1 | . | . | . | 1 |
| ASAL | 2 | . | . | . | . | 2 |
| BD Profound | 8 | | | | | 8 |
| CPT-2 | 3 | . | . | . | . | 3 |
| CTD/CUD | 8 | 8 | 4 | . | . | 20 |
| Cit:type_I | 3 | 5 | 2 | . | . | 10 |
| Cit:type_II | | 1 | . | . | . | 1 |
| GA-1 | 10 | 6 | 3 | . | . | 19 |
| GA-2/MAD | 3 | 1 | 1 | . | . | 5 |
| HCY | 1 | . | . | . | . | 1 |
| IBDHD | 6 | 1 | 1 | . | . | 8 |
| IVA | 6 | 6 | 3 | . | . | 15 |
| LCHADD | 1 | 1 | . | . | . | 2 |
| MAT | 2 | 4 | . | . | . | 6 |
| MCADD | 40 | 29 | 4 | . | . | 73 |
| MCD | | 2 | | | | 2 |

Count of MCAPS Entered in SIS by Disorder Type & Age of Child (8/1/2007 – 2/9/2009)

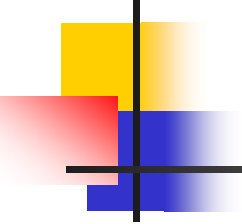
| | 1 st Year | 2 nd Year | 3 rd Year | 4 th Year | 5 th _Year | All |
|--------------------------|----------------------|----------------------|----------------------|----------------------|-----------------------|------------|
| MMA:Cbl AB | 1 | 2 | . | . | . | 3 |
| MMA:Cbl CDF | 10 | 7 | 2 | . | . | 19 |
| MMA:mut- | 9 | 3 | 2 | . | . | 14 |
| MMA:mut0 | 4 | 5 | 1 | . | . | 10 |
| MSUD | 8 | 3 | 2 | . | . | 13 |
| MA (Malonic Aciduria) | 1 | | | | | 1 |
| PKU | 19 | 24 | 22 | 26 | 28 | 119 |
| PKU Variant | 15 | 13 | 11 | 15 | 6 | 60 |
| Prolinemia_Type_I | . | 2 | . | . | . | 2 |
| Prolinemia_Type_II | | 1 | | | | 1 |
| Propionic_acidemia | 2 | 1 | . | . | . | 3 |
| SCADD | 21 | 16 | 5 | . | . | 43 |
| VLADD | 11 | 8 | 2 | . | . | 21 |
| Total (to date) * | 234 | 175 | 71 | 42 | 35 | 557 |

* Note: Counts of all diagnosed disorder types are not displayed

MCAPS Summary

Patient Clinic Status: All Disorders Combined

(8/1/2007 – 2/9/2009)



| | 1st Year (n=234) | 2nd Year (n=175) | 3rd Year (n=71) | 4th Year (n=42) | 5th Year (n=35) |
|--------------------------------|--|--|---------------------------------------|---------------------------------------|---------------------------------------|
| Active Patient | 84% | 80% | 86% | 83% | 86% |
| Lost To Follow-up | 4% | 5% | 4% | 5% | - |
| Refused Treatment | 3% | <1% | - | - | 3% |
| Treatment Deemed Not Necessary | 4% | 7% | 6% | 7% | 3% |
| Patient Died* | 2% | <1% | - | - | - |
| Moved out of state | 1% | 5% | 3% | 2% | 3% |
| Transferred to another center | 2% | 1% | 1% | 2% | 6% |

MCAPS Summary

Developmental Milestones: All Disorders Combined

(8/1/2007 – 2/9/2009)

| | Age Appropriate | | | | |
|-------------------|-----------------|----------|----------------------|----------------|---------------|
| | Speech | Physical | Mental/ Cognitive | Gross Motor | Fine Motor |
| Year 1 (n=234) | 81.2% | 84.2% | 82.5% | 81.6% | 80.3% |
| Year 2 (n=175) | 69.7% | 76.0% | 75.4% | 76.0% | 76.6% |
| Year 3 (n=71) | 69.0% | 81.7% | 76.1% | 83.1% | 77.5% |
| Year 4 (n=42) | 81.0% | 85.7% | 80.1% | 88.1% | 88.1% |
| Year 5 (n=35) | 85.7% | 94.3% | 91.4% | 94.3% | 97.1% |

MCAPS Summary

Developmental Milestones: All Disorders Combined

(8/1/2007 – 2/9/2009)

| | Moderate and/or Severe Delay | | | | |
|-------------------|------------------------------|----------|----------------------|----------------|---------------|
| | Speech | Physical | Mental/ Cognitive | Gross Motor | Fine Motor |
| Year 1 (n=234) | 3.9% | 2.6% | 2.6% | 5.1% | 4.3% |
| Year 2 (n=175) | 7.4% | 3.4% | 4.6% | 5.7% | 5.2% |
| Year 3 (n=71) | 8.4% | 4.2% | 6.0% | 2.8% | 1.4% |
| Year 4 (n=42) | - | - | - | - | - |
| Year 5 (n=35) | 2.9% | - | - | - | - |

MCAPS Summary

Other Outcome Measures: All Disorders Combined

(8/1/2007 – 2/9/2009)

| | Symptoms associated with disorder | Loss of Previous Skills | No Hospitalizations Reported |
|-------------------|-----------------------------------|-------------------------|------------------------------|
| Year 1 (n=234) | 25.2% | 3.0% | 70.9% |
| Year 2 (n=175) | 28.6% | 1.8% | 80.0% |
| Year 3 (n=71) | 19.7% | - | 90.1% |
| Year 4 (n=42) | 14.3% | - | 100% |
| Year 5 (n=35) | 14.3% | - | 100% |

MCAPS Summary

Disorders With Highest Health Services Utilization

(8/1/2007 – 2/9/2009)

| Utilization Measures | Year One | Year Two | Year Three |
|-------------------------|--------------------------------------|-------------------------------------|---|
| Avg. Days Hospitalized | 66 days - PA 27.5 days - MMA:mut0 | 27.2 days - MMA:mut0 6 days - PA | 12.5 days - MMA:Cbl CDF; 12 days - MMA:mut0 |
| Avg. # Hospitalizations | 5 - Cit:type I 3 - MMA:mut0 | 3 - ARG 2 - PA; MSUD | 4 - MMA:mut0; 3 - MMA:mut-; MSUD |
| Avg. # ER Visits | 5 - Cit:type I 2 - MMA:mut0; MSUD | 13 - VLCADD 2 - CIT:type I; ARG | 4 - MMA:mut-; MMA:mut0 1 - VLCADD; MMA:Cbl CDF MSUD; Cit-type I |
| Avg. # Clinic Visits | 11 - MA 11 - MMA:mut0 | 8 - LCHADD 6 - PA; Cit:type I | 5 - Cit-type I 4 - MMA:mut-; MMA:Cbl CDF |

MCAPS Summary

Provider Health Assessment of Child's Health Status

(8/1/2007 – 2/9/2009)

| | Critical | Poor | Fair | Good | Very Good | Excellent |
|-------------------|----------|-------|-------|-------|-----------|-----------|
| Year 1 (n=234) | 9.0% | 9.0% | 6.8% | 18.8% | 32.5% | 23.9% |
| Year 2 (n=175) | 9.7% | 12.6% | 10.3% | 16.0% | 26.9% | 24.6% |
| Year 3 (n=71) | 4.2% | 14.1% | 8.5% | 14.1% | 32.4% | 26.8% |
| Year 4 (n=42) | 11.9% | 7.1% | 2.4% | 7.1% | 19.1% | 52.4% |
| Year 5 (n=35) | 14.3% | 2.9% | 2.9% | 5.7% | 31.4% | 42.9% |

Health Impacts With & Without Screening: MCADD

“...in the absence of newborn screening for MCADD, premature death or serious disability occurs in 20% to 25% of children with the disorder”. (Grosse, Khoury, Greene, Crider, & Pollitt, 2006)

“...33% of survivors will have irreversible neurologic damage”
(Iafolla, Thompson, & Roe, 1994; Pollitt & Leonard, 1998).

| MCADD | Year One (n=40) | Year Two (n=32) |
|------------------------------------|--------------------------|--------------------------|
| In active care at follow-up center | 92.5% | 81.3% |
| Reported deaths* | 0% | 0% |
| No disorder related symptoms | 67.5% | 53.1% |
| Age appropriate cognitive function | 97.5% | 84.4%/3.1% mild |
| # Clinic Visits | 0-2 = 37.5% 3-5 = 45% | 0-2 = 71.9% 3-4 = 25% |
| # Hospitalizations | 0-1 = 90% 2-3 = 10% | 0-1 = 90.6% 2 = 9.4% |

* 3 additional MCADD cases died before follow-up was initiated



Conclusion

This preliminary review demonstrates the feasibility and utility of the California Long Term Follow Up Data System using Metabolic Center Annual Patient Summaries

The surveillance approach addresses:

- Availability of ongoing care and management
- Clinical outcomes and developmental assessment
- Impact on health care utilization



Acknowledgements

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 - Harbor/UCLA Medical Center
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- Kaiser Permanente- Southern California
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- Sutter Memorial Hospital, Sacramento
 - UC Davis Medical Center
 - UC San Francisco Medical Center
 - UCLA Medical Center