Healthcare Service Utilization for Children with Metabolic Disorders Diagnosed Through California Newborn Screening

L. Feuchtbaum, J. Carter, F. Lorey, Genetic Disease Screening Program, California Department of Public Health, Richmond, CA
Background
California Follow-up System (SIS)

- Short-term follow-up for all screened positive cases:
  - Service Report data system to capture interval data covering the case referral, case management and confirmatory testing process by which a diagnosis is confirmed or ruled-out.

- Long-term follow-up for all diagnosed cases:
  - Annual Patient Summaries to track on-going case management and treatment for children through age 5
Long Term Follow-up Using Annual Patient Summaries

- Provides an aggregate profile of similar (rare) disorders, at similar ages, for the first five years of life.
- Build knowledge base describing the full spectrum of clinical outcomes associated with each disorder.
- Impact of newborn screening on the public’s health:
  - Service utilization
  - Access to care
  - Health status
Long Term Follow-up Using Annual Patient Summaries

- Data captured (but not reviewed today)
  - Changes in diagnosis/new genotype data
  - Caregiver adherence/compliance with care
  - Anthropometric measurements to assess growth
  - Type of health care services provided
  - Symptoms/health problems/complications
  - Disorder related interventions
  - Clinician assessment of the development status of the child (cognitive, speech, physical growth, social emotional, gross and fine motor skills)
Long Term Follow-up Using Annual Patient Summaries

- Data captured (and reviewed today)
  - Follow-up status - is child still in care and if not, why?
  - Visits to follow-up care centers
  - Hospitalizations
  - Length of stay
  - Emergency department visits
  - Referrals for additional services
Total responses

- Between July 2005-October 2011:
  - 1,256 metabolic cases detected through NBS
- Since July 2007:
  - 2,280 MCAPS collected (1,082 individuals)
## Patient Follow-up Status

<table>
<thead>
<tr>
<th>Year</th>
<th>Active – patient being seen at center</th>
<th>Follow-up not deemed necessary</th>
<th>Lost to follow-up</th>
<th>Moved out of state</th>
<th>Patient died</th>
<th>Patient not seen</th>
<th>Refused follow-up</th>
<th>Patient transferred to another center</th>
</tr>
</thead>
<tbody>
<tr>
<td>Year 1</td>
<td>81.6%</td>
<td>5.4%</td>
<td>4.5%</td>
<td>2.5%</td>
<td>0.8%</td>
<td>0.3%</td>
<td>3.1%</td>
<td>1.8%</td>
</tr>
<tr>
<td>Year 2</td>
<td>80.1%</td>
<td>6.6%</td>
<td>5.6%</td>
<td>2.8%</td>
<td>0.3%</td>
<td>1.7%</td>
<td>1.8%</td>
<td>1.0%</td>
</tr>
<tr>
<td>Year 3</td>
<td>81.7%</td>
<td>4.8%</td>
<td>5.7%</td>
<td>2.4%</td>
<td>0.5%</td>
<td>3.1%</td>
<td>0.5%</td>
<td>1.4%</td>
</tr>
<tr>
<td>Year 4</td>
<td>82.2%</td>
<td>6.4%</td>
<td>5.5%</td>
<td>1.2%</td>
<td>0.0%</td>
<td>2.8%</td>
<td>0.3%</td>
<td>1.5%</td>
</tr>
<tr>
<td>Year 5</td>
<td>74.5%</td>
<td>11.8%</td>
<td>3.8%</td>
<td>1.9%</td>
<td>0.0%</td>
<td>3.8%</td>
<td>1.4%</td>
<td>2.8%</td>
</tr>
<tr>
<td>All</td>
<td>80.7%</td>
<td>6.4%</td>
<td>5.1%</td>
<td>2.3%</td>
<td>0.4%</td>
<td>1.8%</td>
<td>1.7%</td>
<td>1.6%</td>
</tr>
</tbody>
</table>
Cumulative Patient Follow-up Status

- As of the most recent report received:
- 66.5% of cases were reported to be under active follow-up with a metabolic center.
- For 145 (13.4%) of cases further follow-up was deemed unnecessary
- 4.9% of cases moved out of state
- Ten cases (<1%) died during the LTFU
- 14.5% had parents that refused follow-up or were otherwise lost to follow-up at various ages.
## Count of Clinic Visits Among Referrals to Metabolic Specialty Follow-up Centers

<table>
<thead>
<tr>
<th>Year</th>
<th>n</th>
<th>25%ile</th>
<th>50%ile</th>
<th>75%ile</th>
<th>95%ile</th>
</tr>
</thead>
<tbody>
<tr>
<td>Year 1</td>
<td>719</td>
<td>2</td>
<td>3</td>
<td>5</td>
<td>10</td>
</tr>
<tr>
<td>Year 2</td>
<td>602</td>
<td>1</td>
<td>2</td>
<td>3</td>
<td>5</td>
</tr>
<tr>
<td>Year 3</td>
<td>421</td>
<td>1</td>
<td>1</td>
<td>2</td>
<td>4</td>
</tr>
<tr>
<td>Year 4</td>
<td>326</td>
<td>1</td>
<td>1</td>
<td>2</td>
<td>4</td>
</tr>
<tr>
<td>Year 5</td>
<td>212</td>
<td>0</td>
<td>1</td>
<td>2</td>
<td>4</td>
</tr>
</tbody>
</table>
Contribution of disorder types to total count of clinic visits

Year 1 | Year 2 | Year 3 | Year 4 | Year 5
---|---|---|---|---
PKU | PKU | PKU | PKU | PKU
MCAD deficiency | MCAD deficiency | MCAD deficiency | MCAD deficiency | MCAD deficiency
3MCC | 3MCC | 3MCC | 3MCC | 3MCC
VLCAD deficiency | VLCAD deficiency | VLCAD deficiency | VLCAD deficiency | VLCAD deficiency
GA1 | GA1 | GA1 | GA1 | GA1
MMA cbl CDF | MMA cbl CDF | MMA cbl CDF | MMA cbl CDF | MMA cbl CDF
IVA | IVA | IVA | IVA | IVA
all other disorders | all other disorders | all other disorders | all other disorders | all other disorders
Contribution of disorder types to total count of clinic visits

- Year 1
- Year 2
- Year 3
- Year 4
- Year 5

- all other disorders
- IVA
- MMA cbl CDF
- VLCAD deficiency
- 3MCC
- GA1
- SCAD deficiency
- MCAD deficiency
- PKU
Hospitalizations (n=592)

- 86% No Hospitalizations
- 14% One or more
Contribution of disorder types to total hospitalizations

- MMA cbl CDF
- SCAD deficiency
- CIT Type I
- IVA
- MSUD
- GA1
- MMA mut⁰
- MCAD deficiency

Years:
- Year 1
- Year 2
- Year 3
- Year 4
- Year 5
Contribution of disorder types to total hospitalizations

Year 1: 
- MMA cbl CDF: 20%
- SCAD deficiency: 10%
- CIT Type I: 5%
- IVA: 3%
- MSUD: 2%
- GA1: 1%
- MMA mut⁰: 1%
- MCAD deficiency: 66%
- All other disorders: 7%

Year 2: 
- MMA cbl CDF: 20%
- SCAD deficiency: 10%
- CIT Type I: 5%
- IVA: 3%
- MSUD: 2%
- GA1: 1%
- MMA mut⁰: 1%
- MCAD deficiency: 66%
- All other disorders: 7%

Year 3: 
- MMA cbl CDF: 20%
- SCAD deficiency: 10%
- CIT Type I: 5%
- IVA: 3%
- MSUD: 2%
- GA1: 1%
- MMA mut⁰: 1%
- MCAD deficiency: 66%
- All other disorders: 7%

Year 4: 
- MMA cbl CDF: 20%
- SCAD deficiency: 10%
- CIT Type I: 5%
- IVA: 3%
- MSUD: 2%
- GA1: 1%
- MMA mut⁰: 1%
- MCAD deficiency: 66%
- All other disorders: 7%

Year 5: 
- MMA cbl CDF: 20%
- SCAD deficiency: 10%
- CIT Type I: 5%
- IVA: 3%
- MSUD: 2%
- GA1: 1%
- MMA mut⁰: 1%
- MCAD deficiency: 66%
- All other disorders: 7%
Hospitalizations and Length of Stay

No Hospitalizations

Any Hospitalizations

Total Hospital Days = 3,320

- <7 days (59%)
- 7-30 days (29%)
- >30 days (12%)
Hospital Length of Stay Among Referrals to Metabolic Specialty Follow-up Centers

<table>
<thead>
<tr>
<th>Year</th>
<th>n</th>
<th>25%ile</th>
<th>50%ile</th>
<th>75%ile</th>
<th>95%ile</th>
</tr>
</thead>
<tbody>
<tr>
<td>Year 1</td>
<td>n = 110</td>
<td>4</td>
<td>7.5</td>
<td>26</td>
<td>88</td>
</tr>
<tr>
<td>Year 2</td>
<td>n = 67</td>
<td>2</td>
<td>4</td>
<td>10</td>
<td>20</td>
</tr>
<tr>
<td>Year 3</td>
<td>n = 27</td>
<td>2</td>
<td>3</td>
<td>9</td>
<td>44</td>
</tr>
<tr>
<td>Year 4</td>
<td>n = 19</td>
<td>2</td>
<td>3</td>
<td>4</td>
<td>22</td>
</tr>
<tr>
<td>Year 5</td>
<td>n = 3</td>
<td>3</td>
<td>5</td>
<td>9</td>
<td>9</td>
</tr>
</tbody>
</table>
ER visits (n=490)

- No ER visits: 89%
- One or more: 11%

<table>
<thead>
<tr>
<th>Visits</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>1 visit</td>
<td>63%</td>
</tr>
<tr>
<td>2 visits</td>
<td>18%</td>
</tr>
<tr>
<td>3 visits</td>
<td>8%</td>
</tr>
<tr>
<td>4 visits</td>
<td>6%</td>
</tr>
<tr>
<td>5 or more</td>
<td>5%</td>
</tr>
</tbody>
</table>
Contribution of disorder types to Emergency Department Visits

- All other disorders
- MMA mut⁻
- SCAD deficiency
- CIT Type I
- IVA
- MSUD
- GA1
- MMA mut⁰
- MCAD deficiency

Year 1, Year 2, Year 3, Year 4, Year 5
Contribution of disorder types to Emergency Department Visits

- All other disorders
- MMA mut^-
- SCAD deficiency
- CIT Type I
- MSUD
- GA1
- MMA mut^0
- MCAD deficiency

Year 1
Year 2
Year 3
Year 4
Year 5
Assessments and Referrals for Additional Services Among 726 Cases

<table>
<thead>
<tr>
<th>Type</th>
<th>%</th>
</tr>
</thead>
<tbody>
<tr>
<td>Provider review of milestones</td>
<td>49.6</td>
</tr>
<tr>
<td>Developmental assessment using standardized instrument</td>
<td>4.3</td>
</tr>
<tr>
<td>Referred or receiving ongoing physical/occupational therapy</td>
<td>8.7</td>
</tr>
<tr>
<td>Referred or receiving ongoing speech therapy</td>
<td>6.6</td>
</tr>
</tbody>
</table>


Why does this matter?

- Because more kids are now getting diagnosed with disorders, preventative health service utilization for genetic disorders would be expected to increase.
- Due to earlier diagnosis, the hope is that ER and hospitalization use decreases with better disease management.
- But we won’t really know if this is happening if we don’t follow-up!
Future Steps

- Continue to slice and dice the data to answer public health/clinical outcomes questions
- Improve data quality
  - Better capture hospitalization information – validate with hospital discharge data?
- Find out what issues people want to know most about.....
Acknowledgements

Special thanks to staff at the following State-contracted metabolic follow-up centers who contribute data

- Cedar-Sinai Medical Center
- Children’s Hospital and Research Center-Oakland
  - Children’s Hospital Central California
  - Children’s Hospital Los Angeles
- Children’s Hospital of Orange County
- Children’s Hospital San Diego Health Center
- Harbor/UCLA Medical Center
- Kaiser Permanente-Northern California
- Kaiser Permanente- Southern California
  - LAC/USC Medical Center
- Stanford University Medical Center
- Sutter Memorial Hospital, Sacramento
  - UC Davis Medical Center
- UC San Francisco Medical Center
  - UCLA Medical Center

and CDC Cooperative Agreement #5U50DD000475