LONG-TERM FOLLOW-UP OF CONGENITAL ENDOCRINE DISORDERS DIAGNOSED THROUGH THE CALIFORNIA NEWBORN SCREENING PROGRAM

Lisa Feuchtbaum, DrPH, MPH
Ning Rosenthal, MD, PhD
Genetic Disease Screening Program
California Department of Public Health
Background

• Endocrine screening
  • 1980 for congenital hypothyroidism
  • 2005 for congenital adrenal hyperplasia

• Birth Prevalence in California
  • PCH: 1 in every 1,706
  • CAH: 1 in every 17,241 (~80% Salt wasting)

• July 2005- Implementation of a web-based Screening Information System (SIS)

• August 2009 – Implemented LTFU for endocrine disorders using Annual Patient Summary approach at 7-state contracted endocrine follow-up centers
Long Term Follow-Up Data System

Annual Patient Summaries (APS)

• Provides an aggregated yearly “snapshot” of children diagnosed through NBS from age one through age five

• Monitor availability of ongoing care, health care utilization and developmental outcomes
Number of Case Reports by Disorder and Year

- Total: 139, 102, 67
- Hypothyroidism: 122, 74, 61
- CAH: 17, 10, 8

* as of 3/31/2013
Insurance Coverage: CH & CAH Reports Combined

- Medicaid/CCS/Public Program
- Military/Government/CHAMPUS
- Private insurance/employer sponsored
- Self-pay/uninsured
- Unknown

Bar chart showing the percentage of insurance coverage across different years and types.
Patient Clinic Visits/Year

Mean Number of Visits

1-year | 2-year | 3-year | 4-year | 5-year
---|---|---|---|---
CH | CAH

Data showing the mean number of clinic visits per year for CH and CAH patients.
CAH: Emergency Room Visits & Hospitalizations

Note: Very few ER visits & hospitalizations for CH in any given year
Disorder-Related Symptoms

Congenital Hypothyroidism (CH)

Year-1: 13%
Year-2: 30%
Year-3: 30%
Year-4: 18%
Year-5: 18%

Overall Most Common symptoms:
Developmental Delay: 5.7%
Constipation: 9.7%
Disorder-Related Symptoms

Congenital Adrenal Hyperplasia (CAH)

Year-1: 47%
Year-2: 40%
Year-3: 25%
Year-4: 30%
Year-5: ~

Overall Most common symptom:
Ambiguous genitalia 22.6%
Hypertension: 6%
Vomiting: 6%
Dehydration: 6%
CH: Disorder-Related Assessments

- Bone age radiographic studies
- Hormone level studies
- Thyroid scan/ultrasound
- Other assessment

<table>
<thead>
<tr>
<th>Year</th>
<th>Bone age radiographic studies</th>
<th>Hormone level studies</th>
<th>Thyroid scan/ultrasound</th>
<th>Other assessment</th>
</tr>
</thead>
<tbody>
<tr>
<td>1-year</td>
<td>85</td>
<td>75</td>
<td>5</td>
<td>2</td>
</tr>
<tr>
<td>2-year</td>
<td>80</td>
<td>70</td>
<td>4</td>
<td>3</td>
</tr>
<tr>
<td>3-year</td>
<td>75</td>
<td>65</td>
<td>3</td>
<td>4</td>
</tr>
<tr>
<td>4-year</td>
<td>70</td>
<td>60</td>
<td>2</td>
<td>5</td>
</tr>
<tr>
<td>5-year</td>
<td>65</td>
<td>55</td>
<td>1</td>
<td>6</td>
</tr>
</tbody>
</table>
CAH: Disorder-Related Assessments

- Bone age radiographic studies
- Hormone level studies
- Thyroid scan/ultrasound
- Other assessment

Percent
CH: Treatments Indicated

Year 1  Year 2  Year 3  Year 4  Year 5

<table>
<thead>
<tr>
<th>Treatment Type</th>
<th>Year 1</th>
<th>Year 2</th>
<th>Year 3</th>
<th>Year 4</th>
<th>Year 5</th>
</tr>
</thead>
<tbody>
<tr>
<td>Medication-Thyroxine</td>
<td>100</td>
<td>80</td>
<td>70</td>
<td>60</td>
<td>50</td>
</tr>
<tr>
<td>Other dietary supplements</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>Other treatments</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>Treatment deemed not necessary</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
</tr>
</tbody>
</table>
Loss of Previously Acquired Skills

CH  CAH

Year 1  Year 2  Year 3  Year 4  Year 5
Conclusions

• Data provide a useful snapshot of the service utilization patterns and overall health status of CH and CAH patients

• Only a small percent of children had serious disorder-related complications

• ER visits & hospitalizations are more common among CAH patients compared to CH patients

• Data identified gaps in services that should be provided more frequently: consultations with primary care providers or referrals for genetic counseling services

• Future use of data could examine how treatments/services provided correspond with recommended guidelines for care
Thank you!

Contact information

- Lisa Feuchtbaum: Lisa.Feuchtbaum@cdph.ca.gov

- Special thanks to the state-contracted endocrine centers that provided the annual patient summary data