

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

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# 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

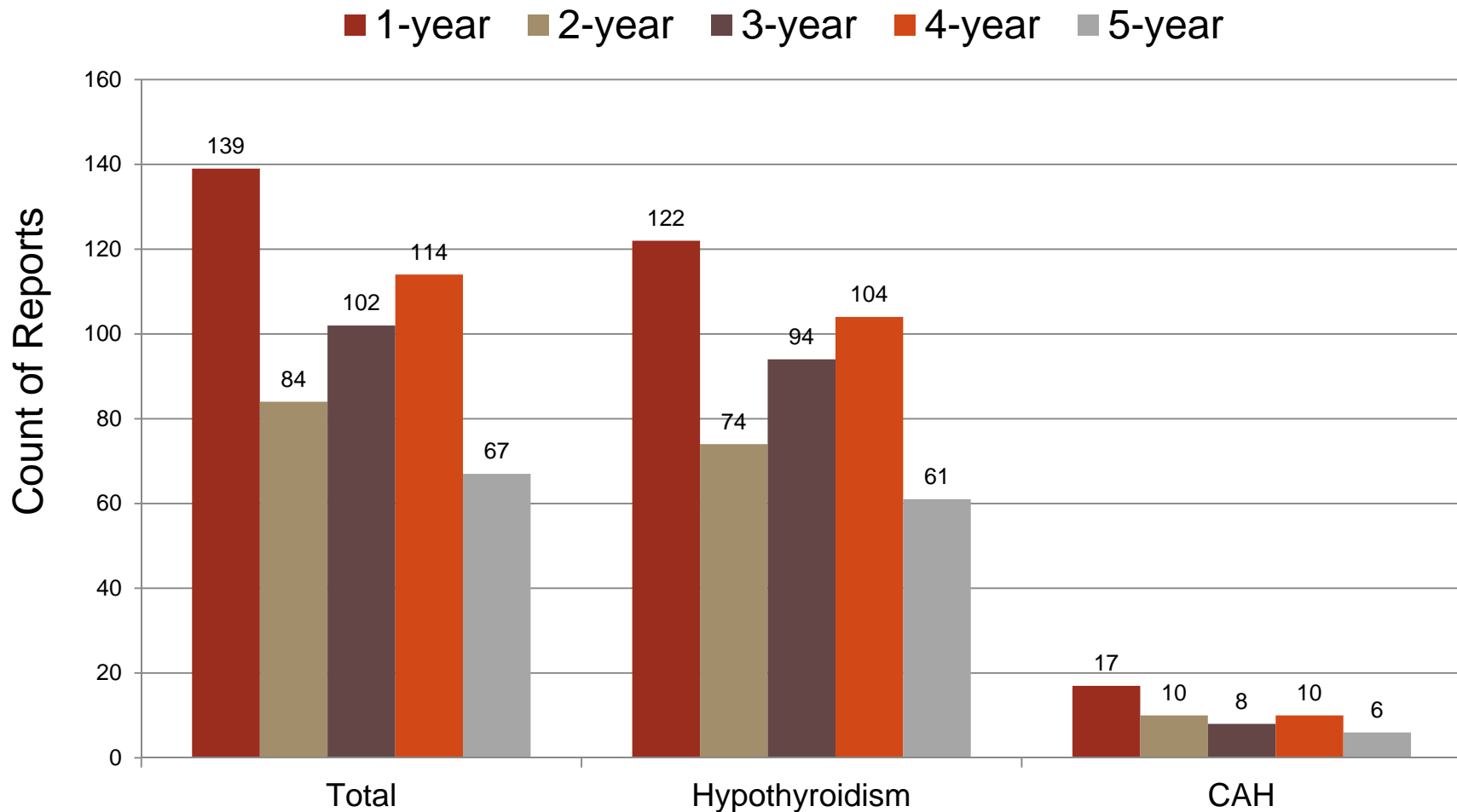


# Long Term Follow-Up Data System

## 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

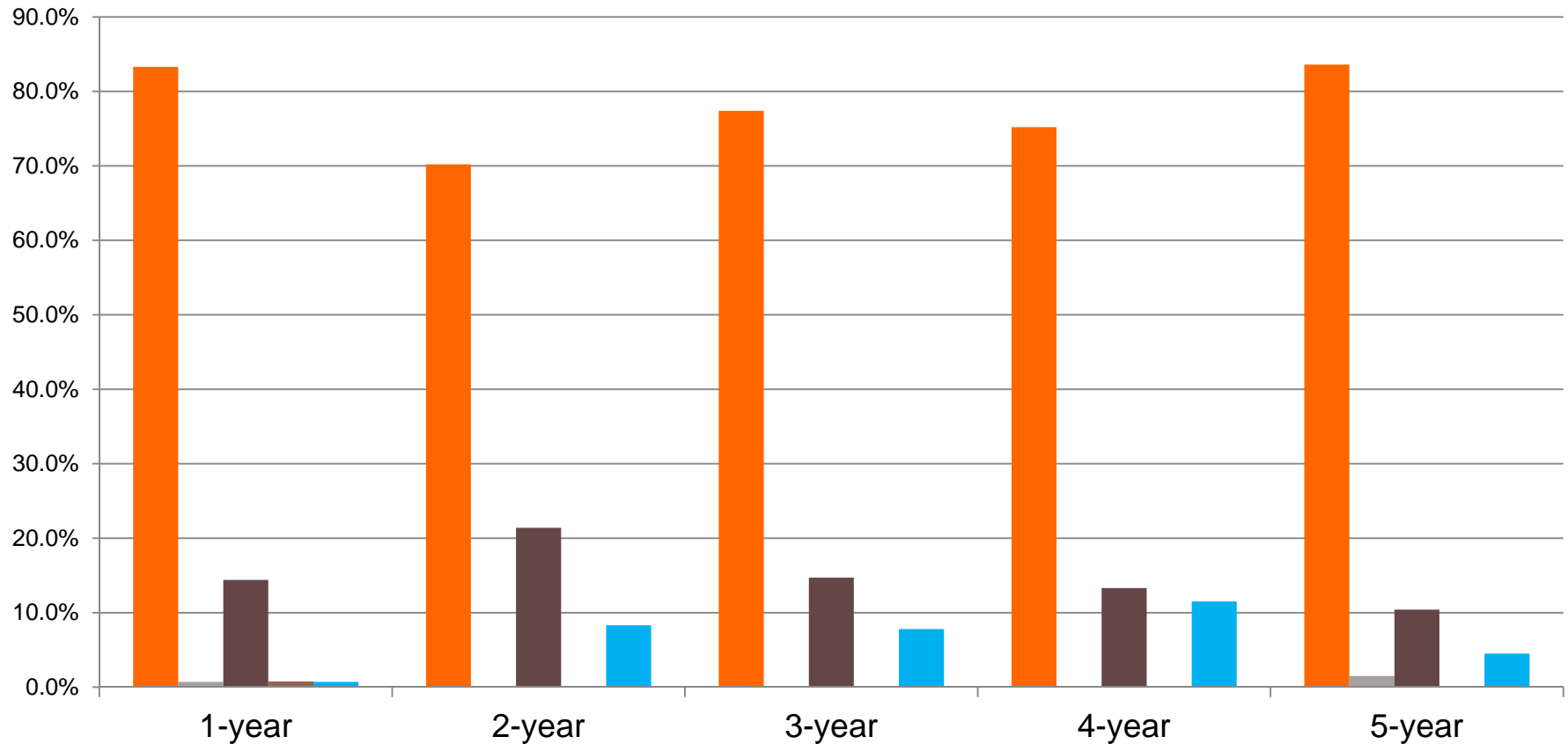
# Number of Case Reports by Disorder and Year



\* as of 3/31/2013

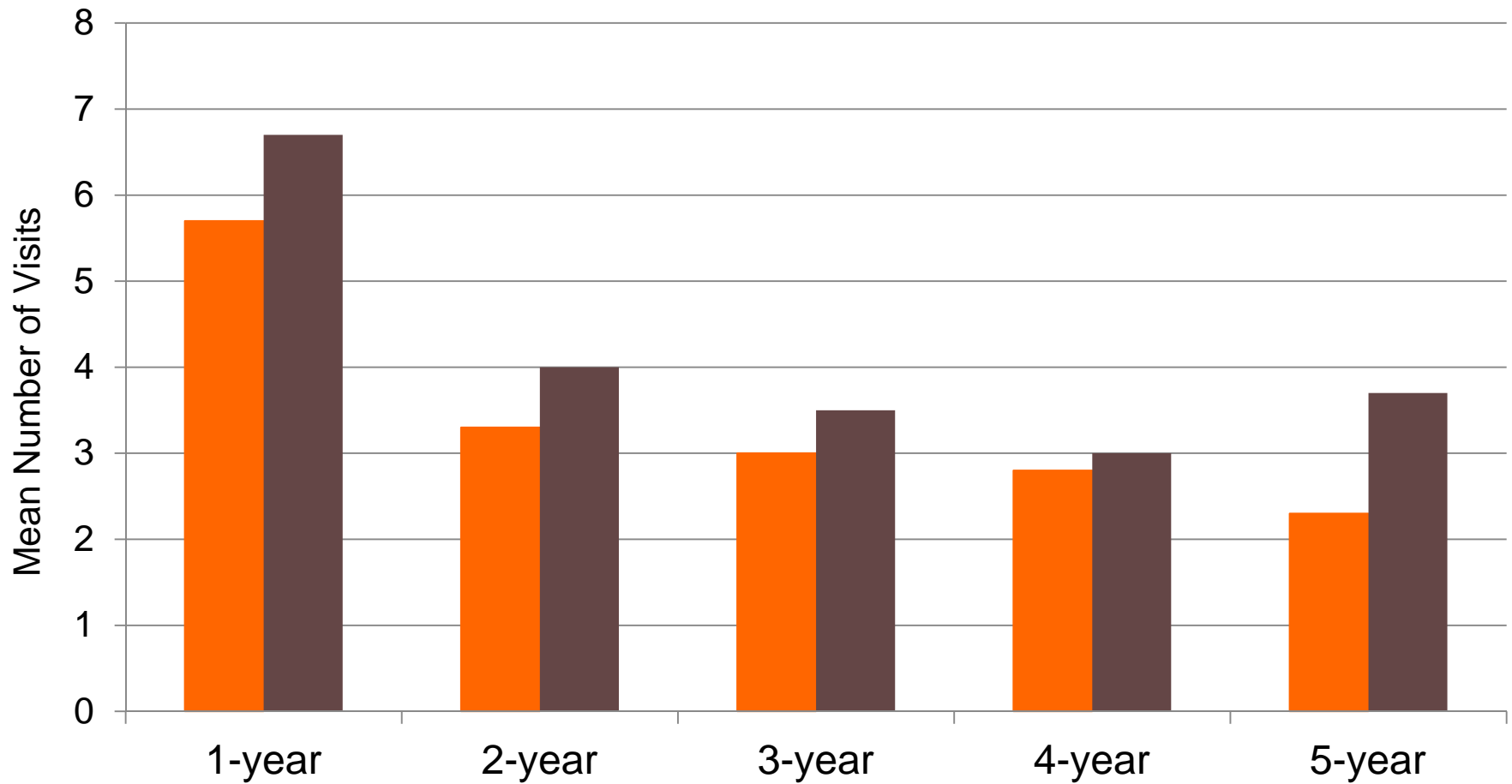
# Insurance Coverage: CH & CAH Reports Combined

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



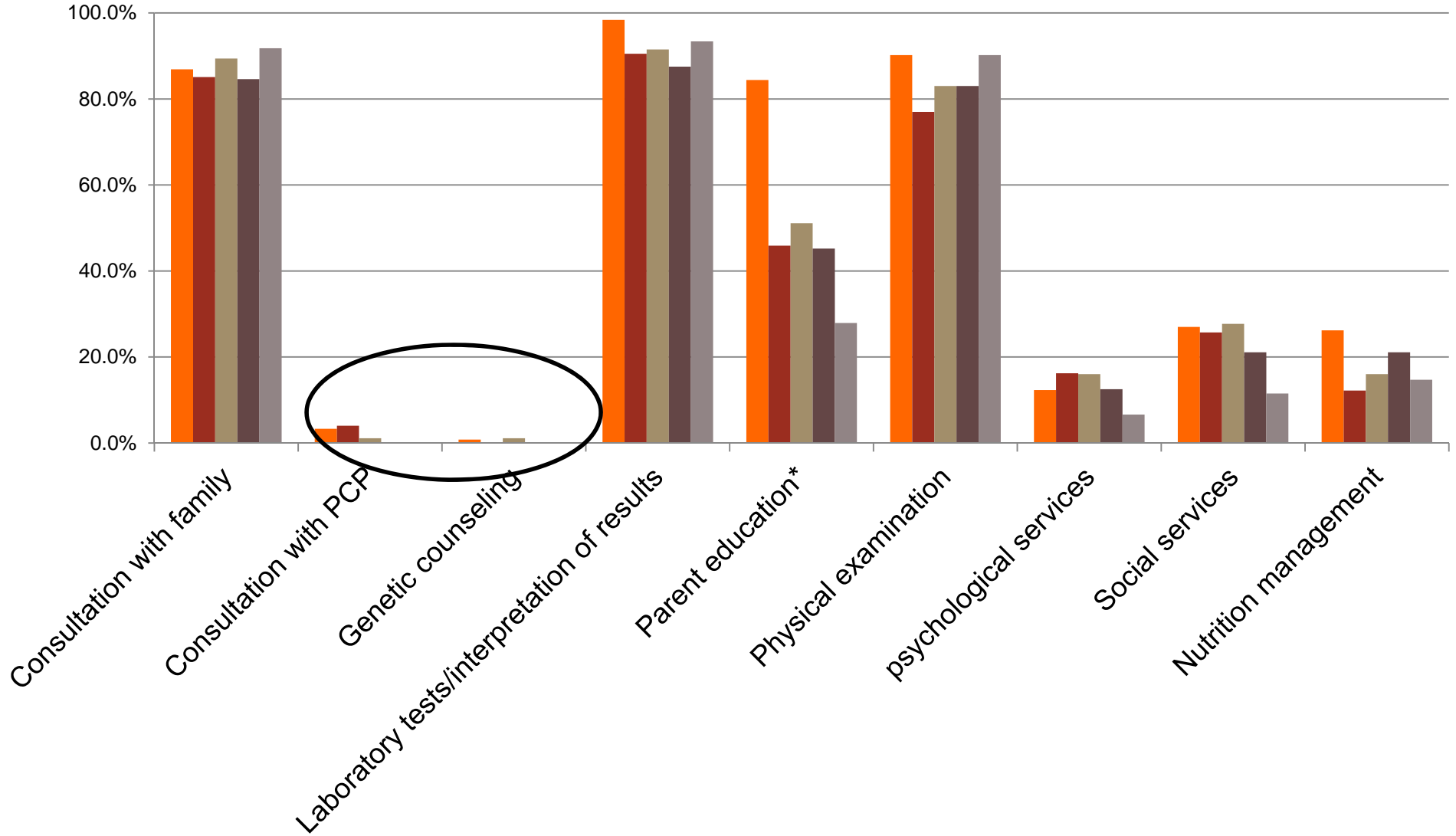
# Patient Clinic Visits/Year

CH CAH



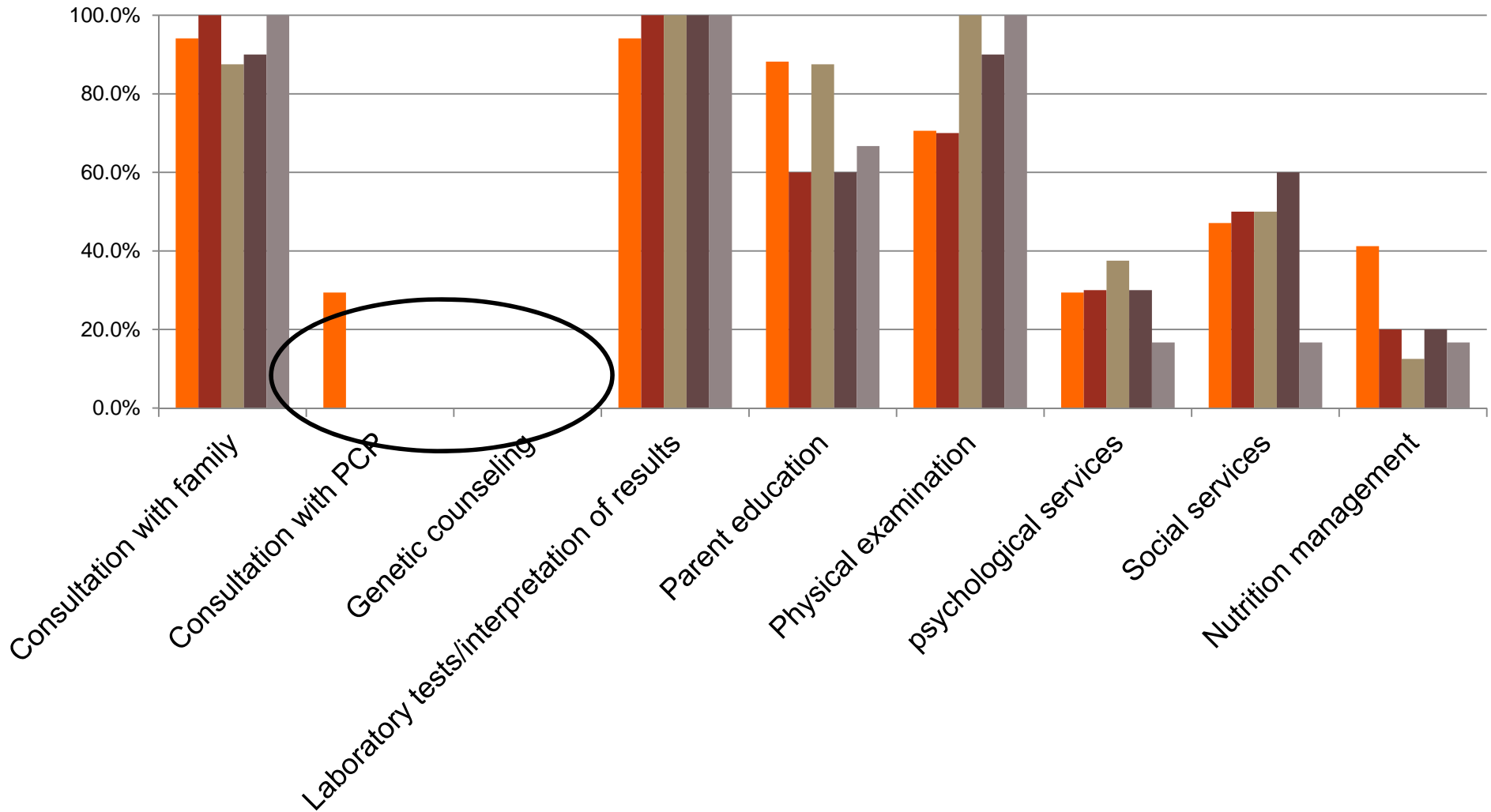
# Services Provided: CH

1-year 2-year 3-year 4-year 5-year



# Services Provided: CAH

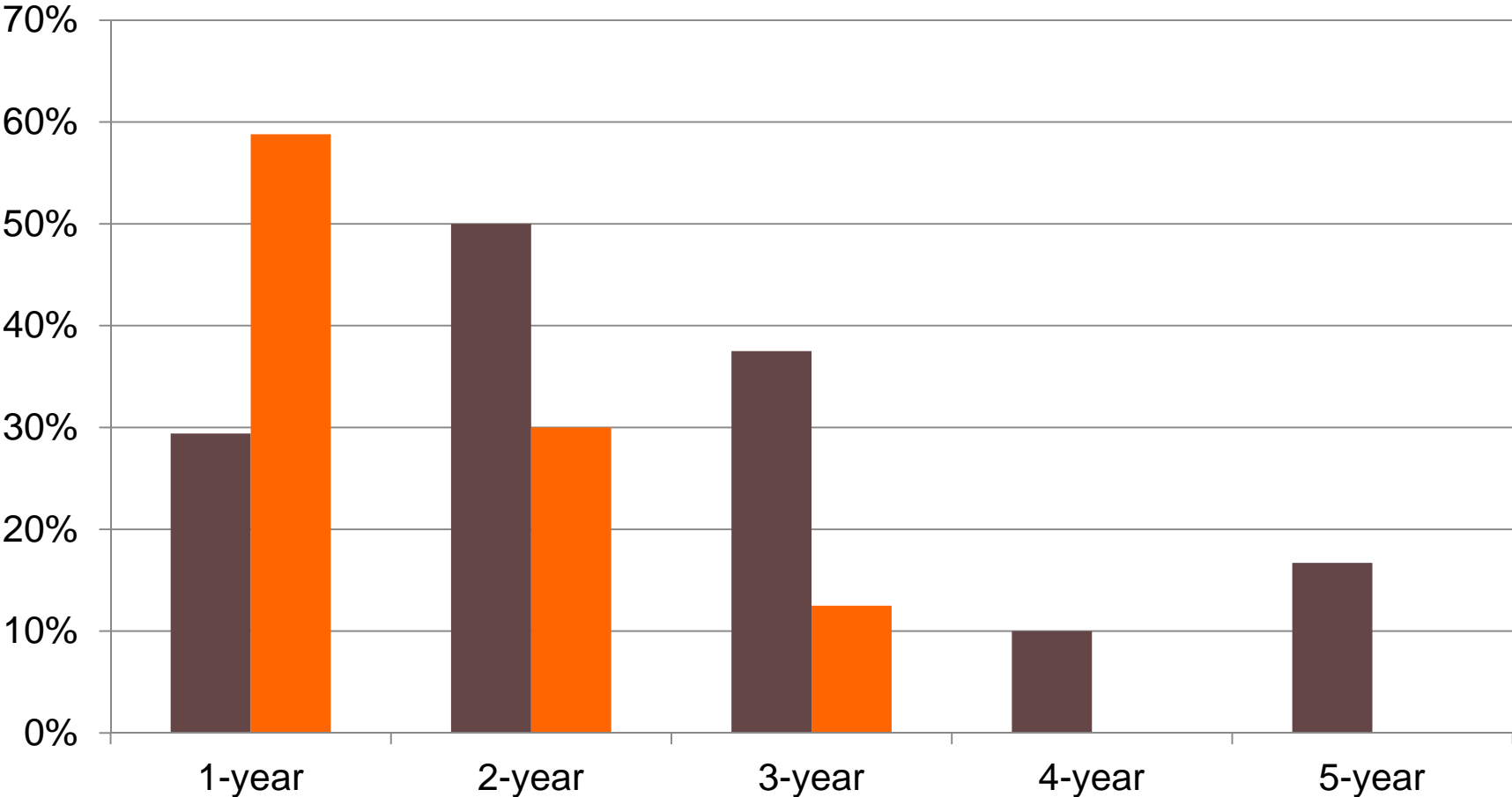
1-year 2-year 3-year 4-year 5-year





# CAH: Emergency Room Visits & Hospitalizations

■ ER 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%

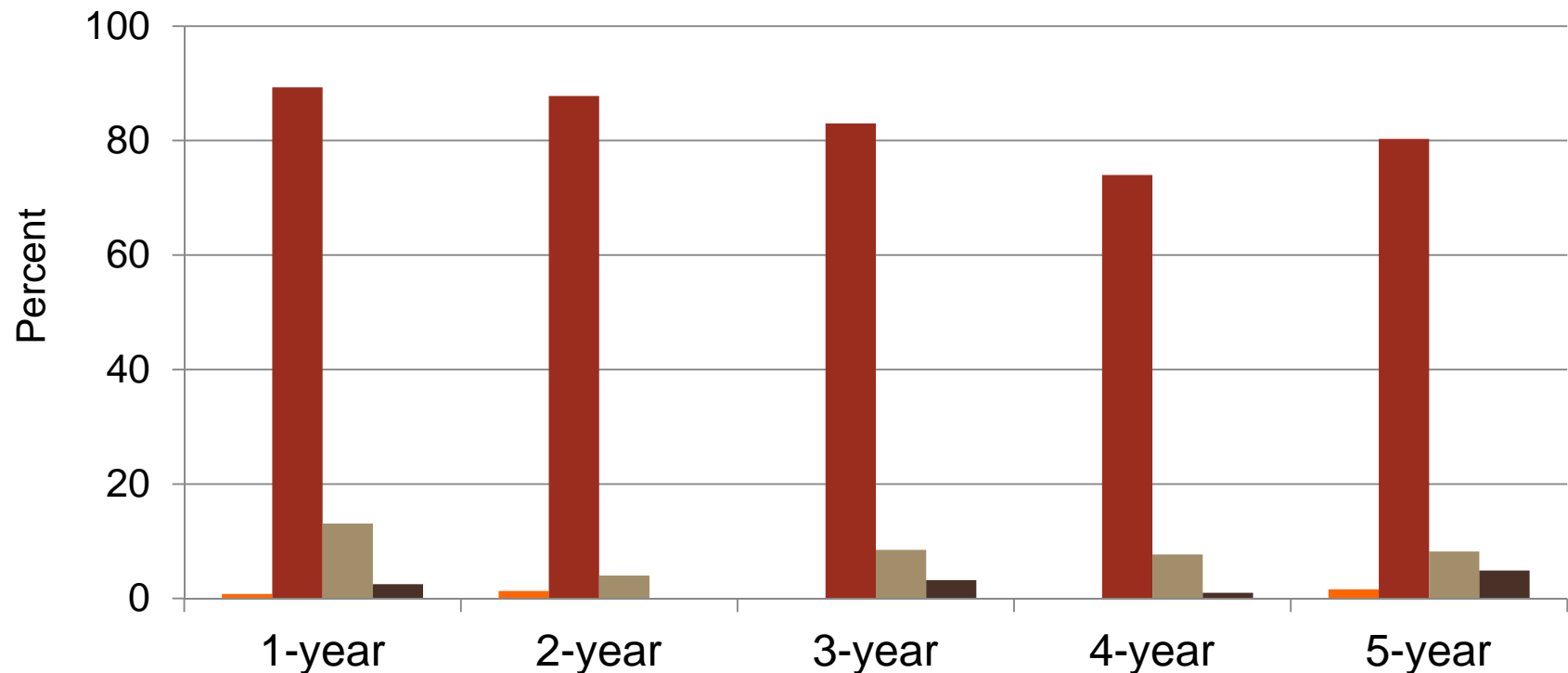
Hypertension: 6%

Vomiting: 6%

Dehydration: 6%

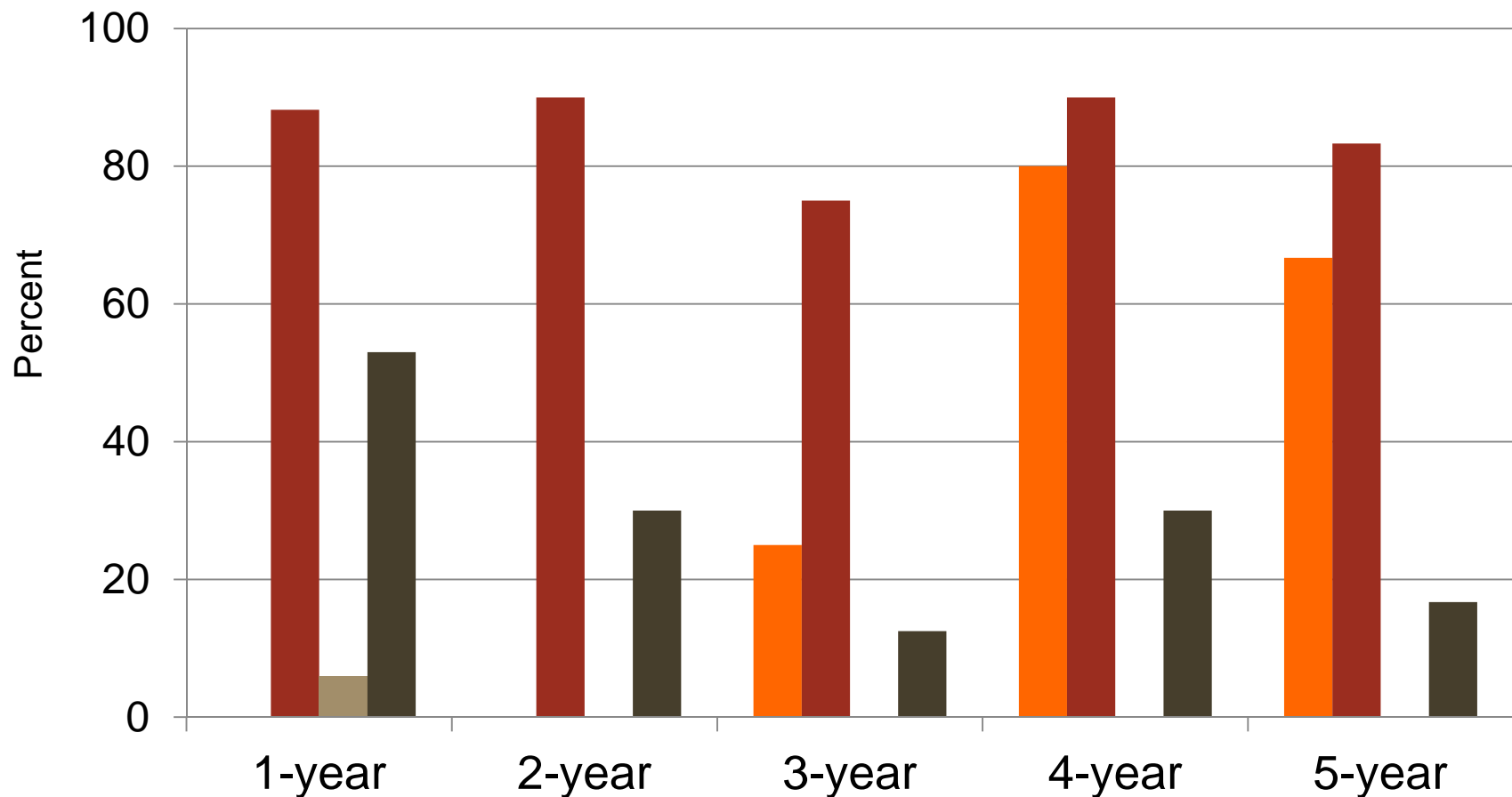
# CH: Disorder-Related Assessments

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



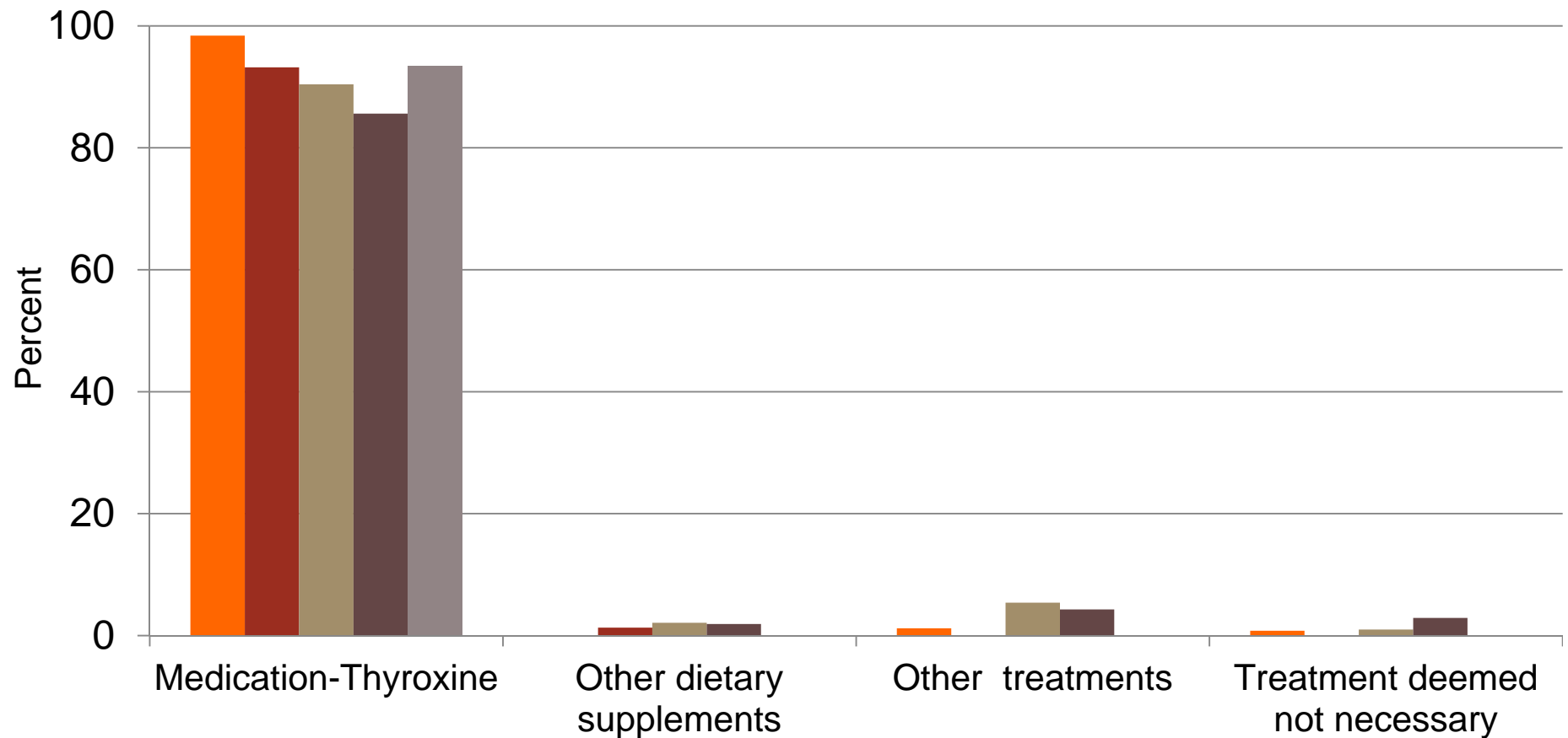
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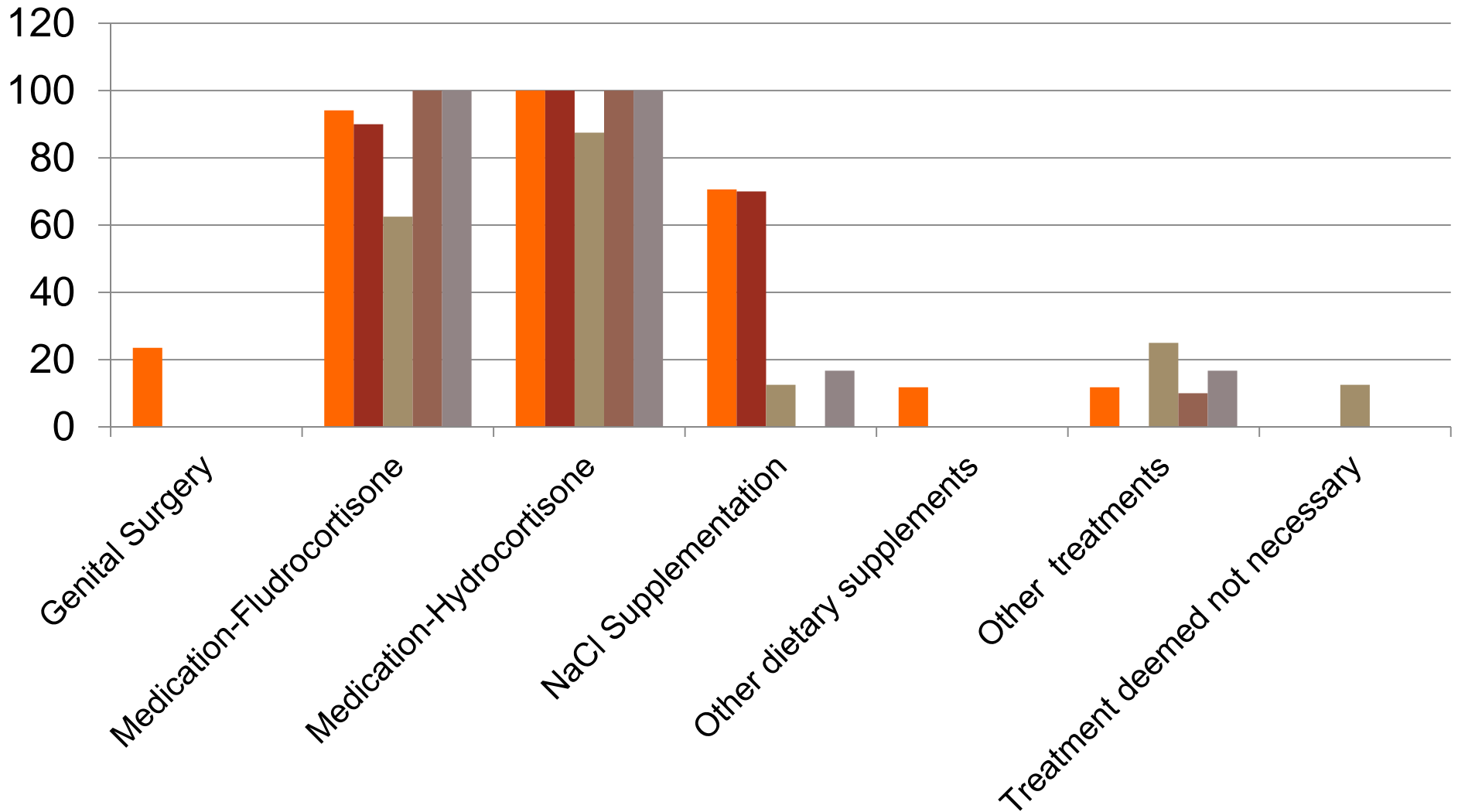
# CH: Treatments Indicated

Year 1 Year 2 Year 3 Year 4 Year 5



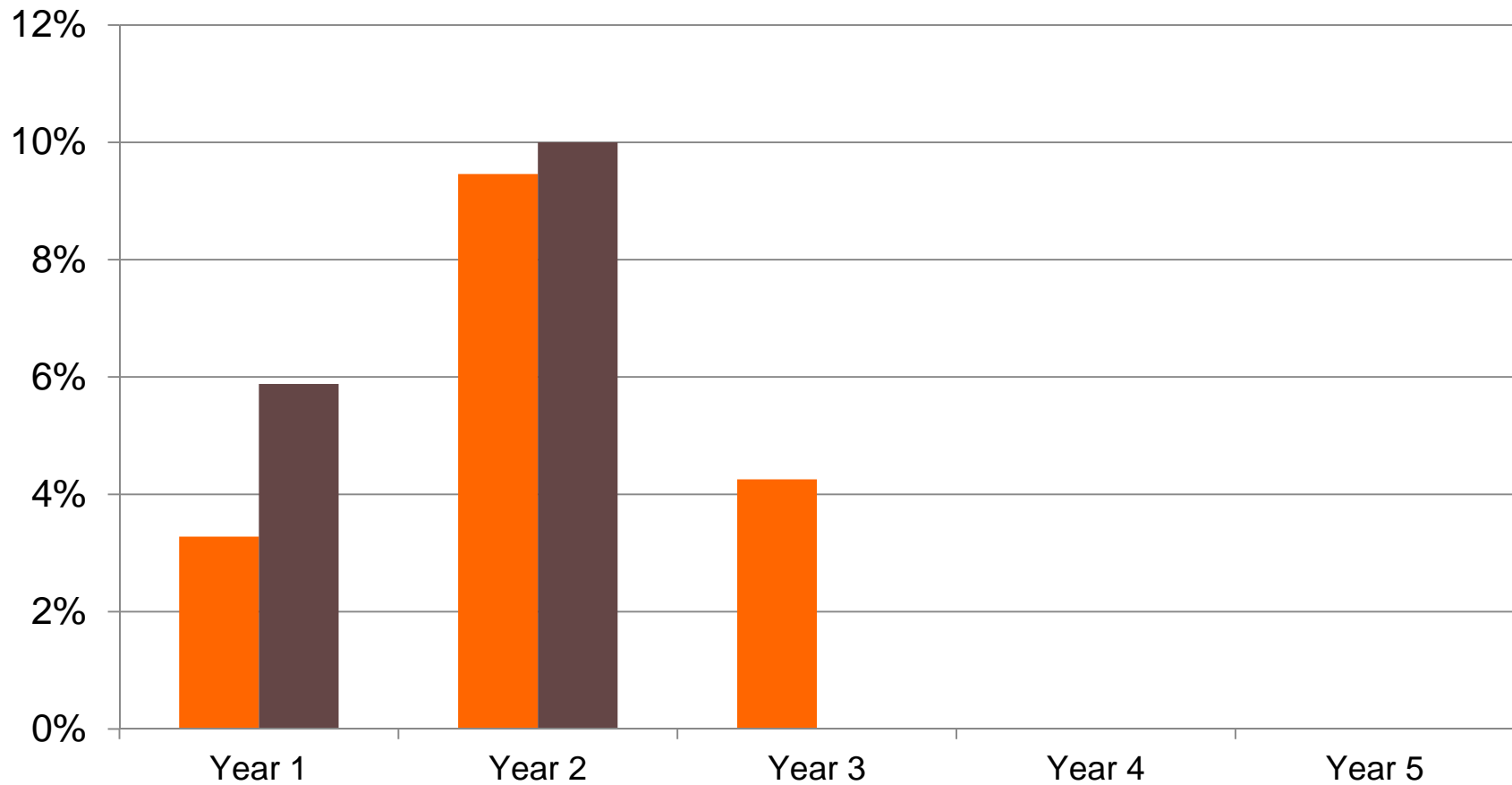
# CAH: Treatments Indicated

Year 1 Year 2 Year 3 Year 4 Year 5



# Loss of Previously Acquired Skills

CH CAH





# 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](mailto:Lisa.Feuchtbaum@cdph.ca.gov)
- Special thanks to the state-contracted endocrine centers that provided the annual patient summary data