

California Newborn Screening Program

Follow-up Status

During First Five Years of Life

for Select Primary RUSP Disorders

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Background: RUSP

- Disorders on the Recommended Uniform Screening Panel (RUSP) have had to meet the criteria that a treatment exists and through early diagnosis, the treatment can halt or slow disease progression.
- Assumes that specialty care follow-up centers provide disease treatment and management services that are accessible to families in the early years of life.



Background:

California Newborn Screening Program (NBS)

- 1980:
 - California NBS Program began.
 - ~500,000 newborns/year.
- 2005:
 - MS/MS screening started.
 - Electronic data collection started (SIS).
 - Long-Term Follow-up (LTFU) data system using Annual Patient Summary (APS) reports started.



California Newborn Screening Program Long-Term Follow Up Approach

Annual Patient Summary (APS) Reports:

- Collected for program evaluation purposes
- Data provided by state-contracted specialty care follow-up centers
- Once a year assessment of status of the child through fifth birthday
- Reports document whether child is still in active care
- Clinical management strategies
- Clinical outcomes



California Newborn Screening Program Follow Up Model

Clinical case coordinators refer screen positive newborns to state-contracted specialty care follow-up centers



Follow-up centers responsible Short Term Follow-Up: documentation of the services provided, health status of newborn & outcomes of confirmatory testing



No Disorder



Confirmed Disorder



Initiation of Long Term Follow-Up via Annual Patient Summary Data Collection (through age 5)



Study Questions

- What percent of children with disorders remain in care between the ages of one and five years old?
- What percent become lost to follow-up?
- What are the characteristics of lost to follow-up?
- What percent died?

Consensus public health questions based on:

(1) Hinton et al paper: What questions should newborn screening long term follow-up be able to answer..." Genetics in Medicine, Vol 13, No 10, Oct 2011

(2) Priority questions determined by Newborn Screening Translational Research Network



Methods

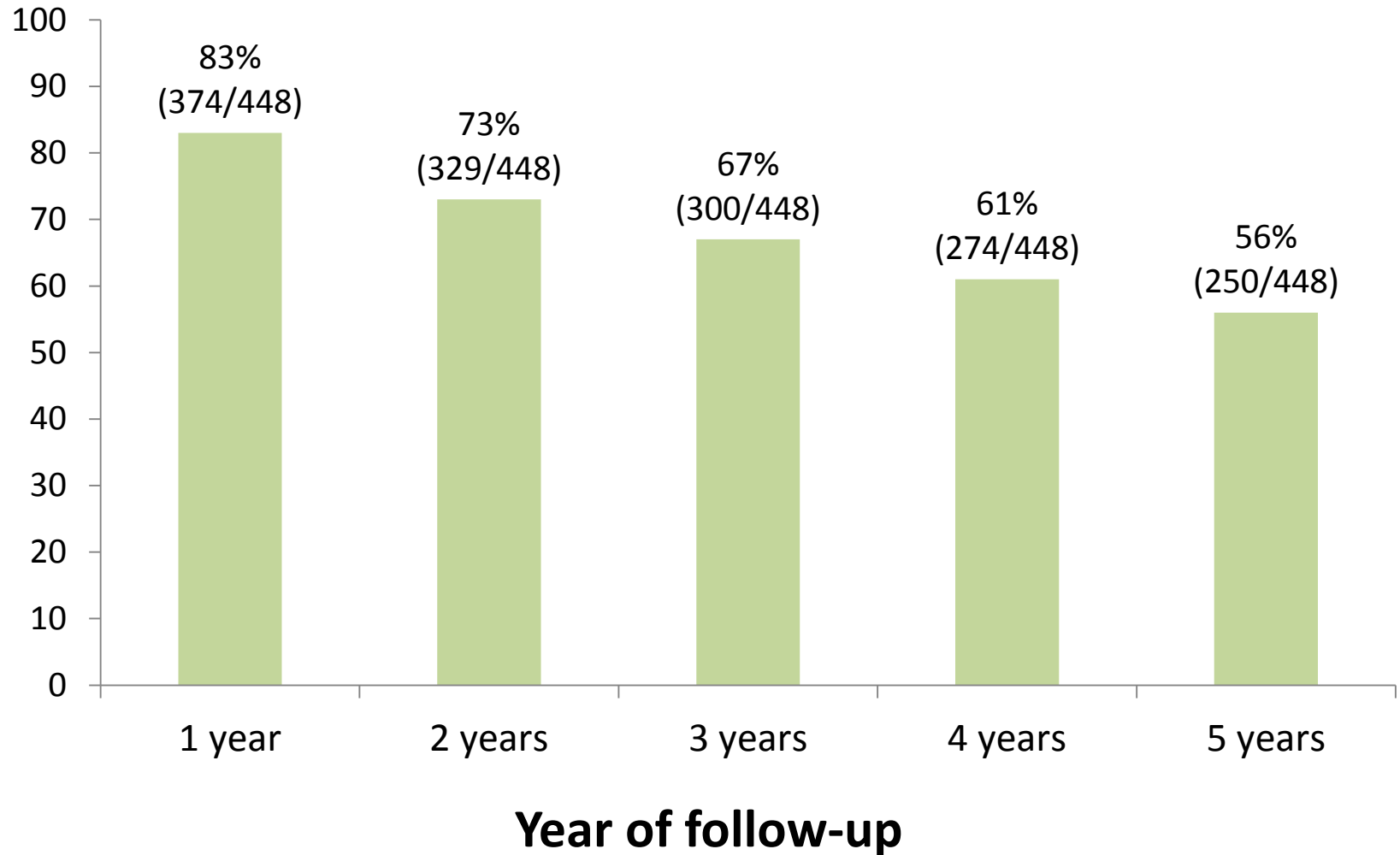
- 19 primary RUSP metabolic disorders
- 448 patients identified between 07/07/2005 through 12/31/2009
- Five years of follow-up data
- For data analysis including:
 - Newborn screening program routine data including metabolic disorder diagnosis, maternal race-ethnicity, and maternal age
 - APS data including follow-up status and child age at APS report
 - Birth certificate data including maternal education, and payment for delivery



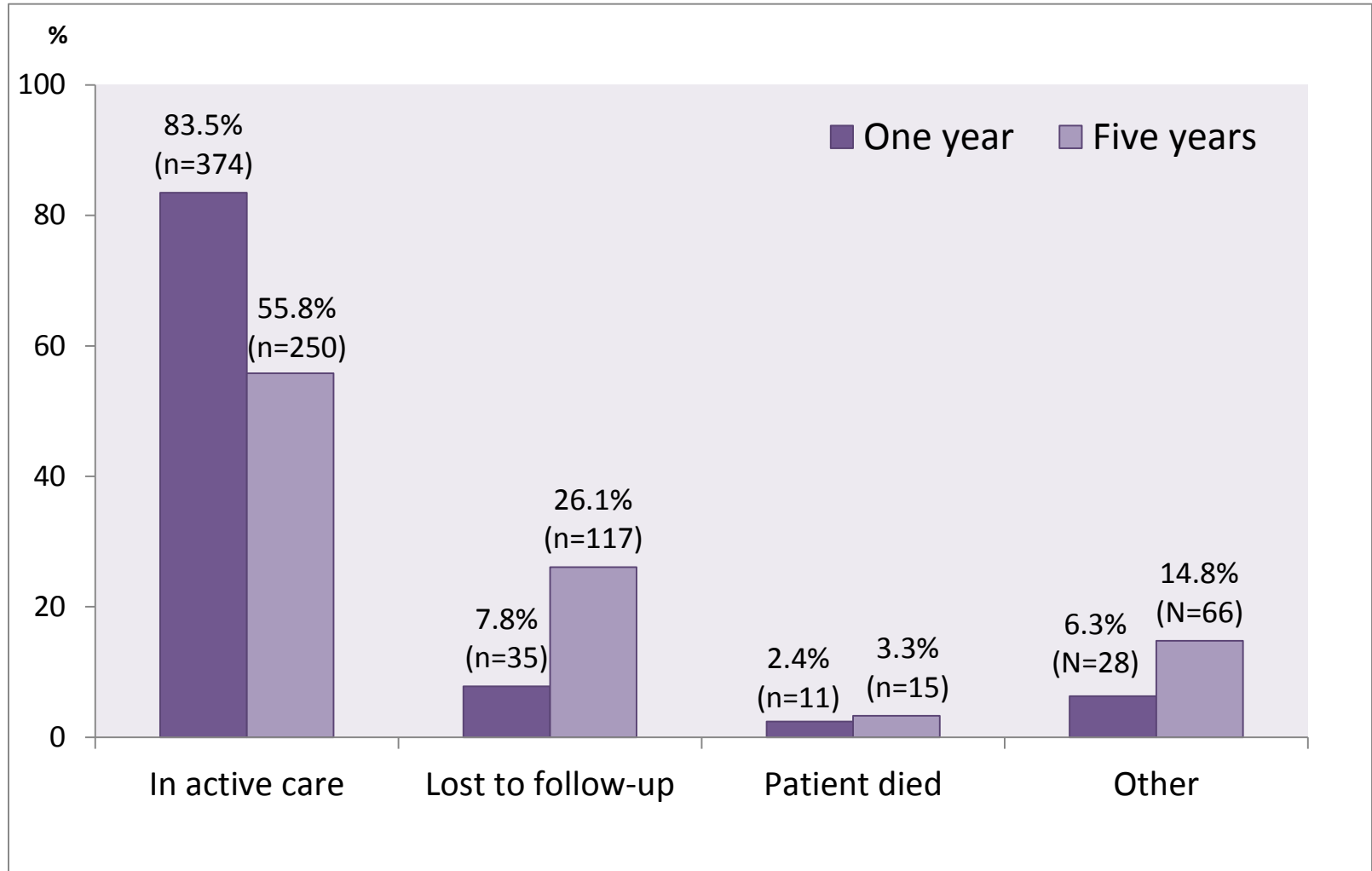
Methods

- Follow-up categories on APS report:
 - Active Care: active, transferred to another center, followed up but no in-person visit
 - Lost to Follow-Up: lost to follow-up, refused follow-up
 - Other: child died, moved out of state, treatment deemed not necessary
- Assign follow-up status to missed APS reports
 - Assign 'Active' to a missed APS report if it is followed by an 'Active' APS in later years.
 - Assign 'lost to follow-up' if there are no more subsequent APS reports with patient visit.
- Received 1,590 APS reports for the 448 patients over five-year follow-up.

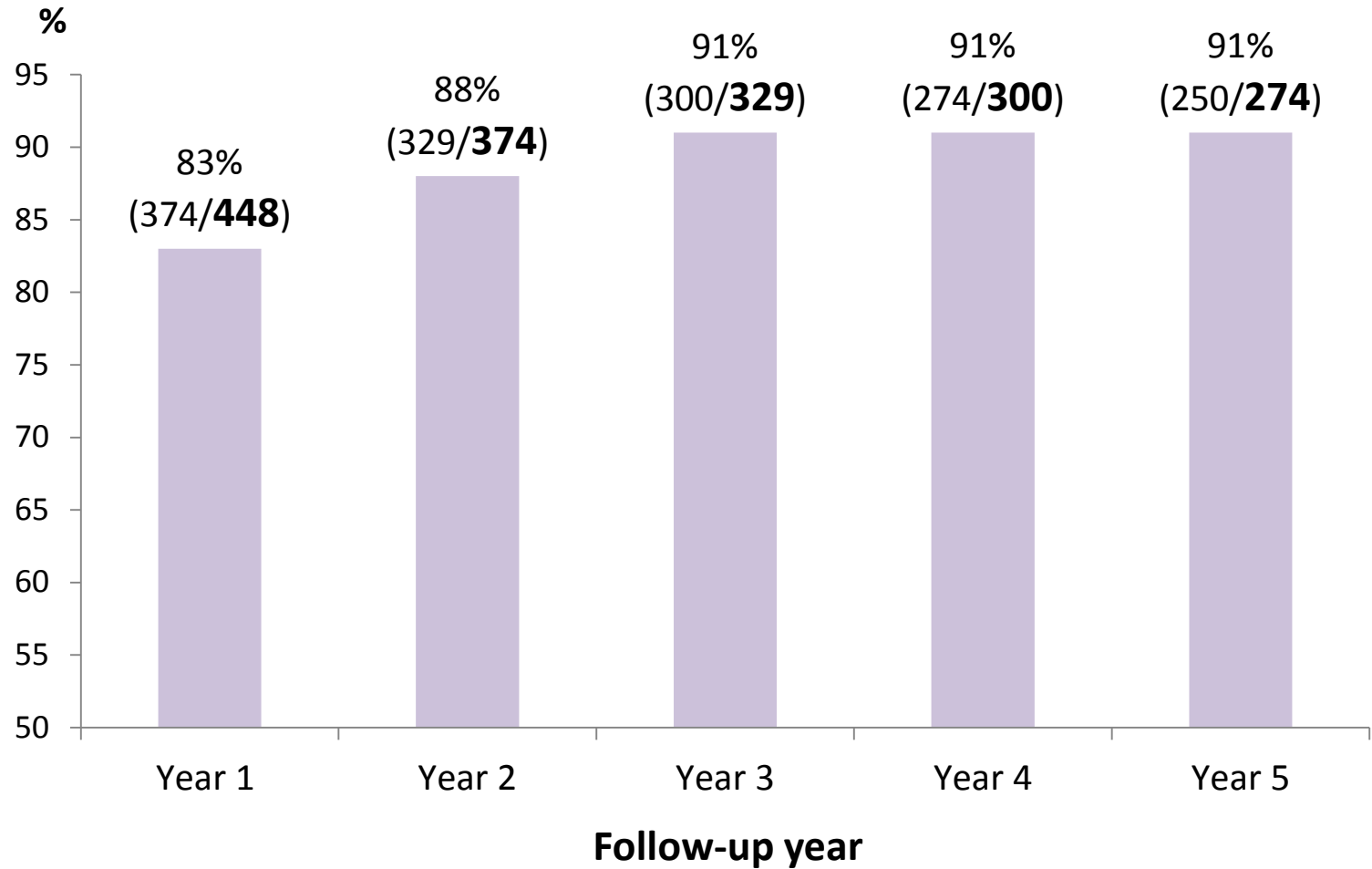
Cumulative % of initial cohort remaining in active care by follow-up year (n=488)



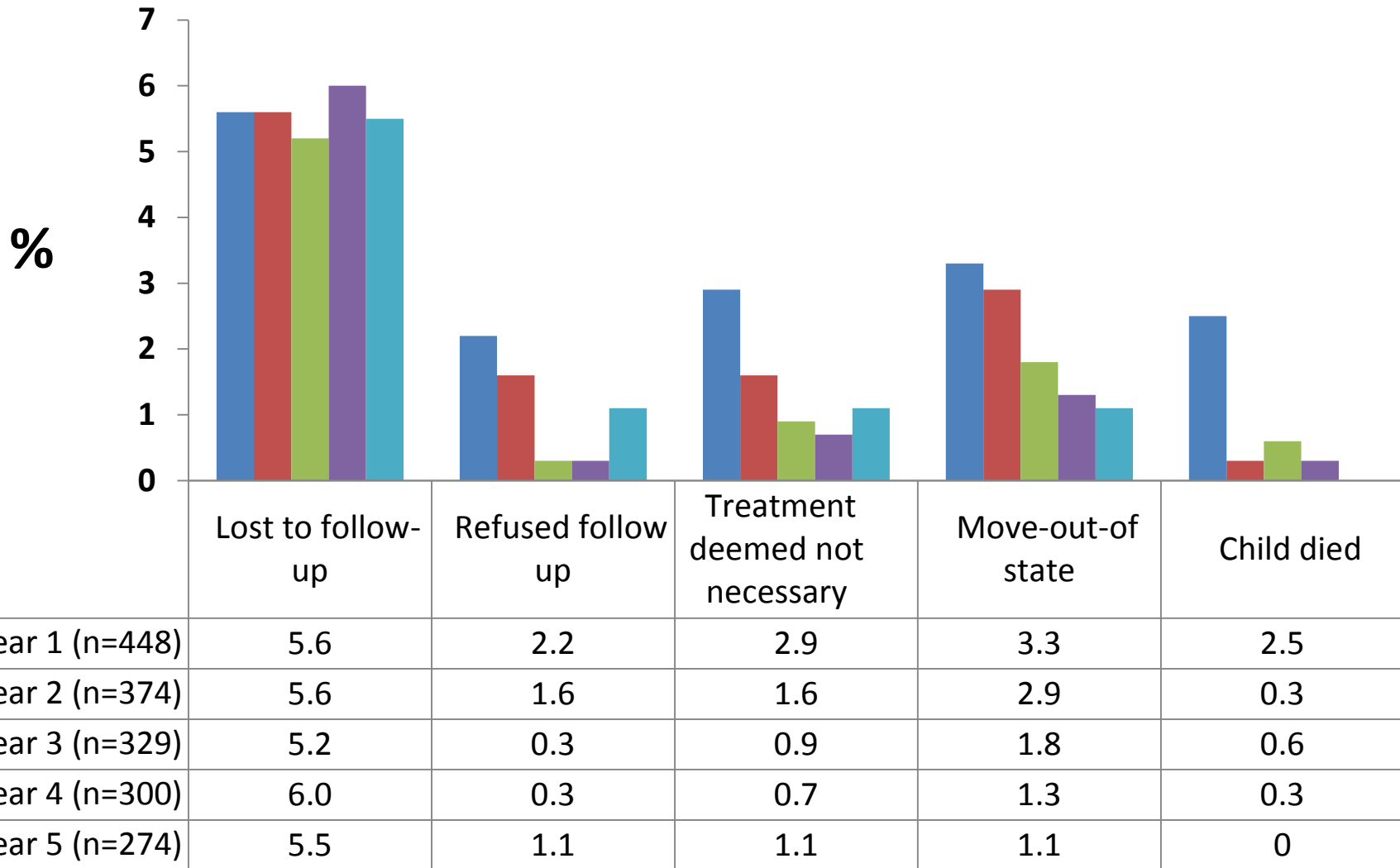
Comparison of one-year and five-year follow-up



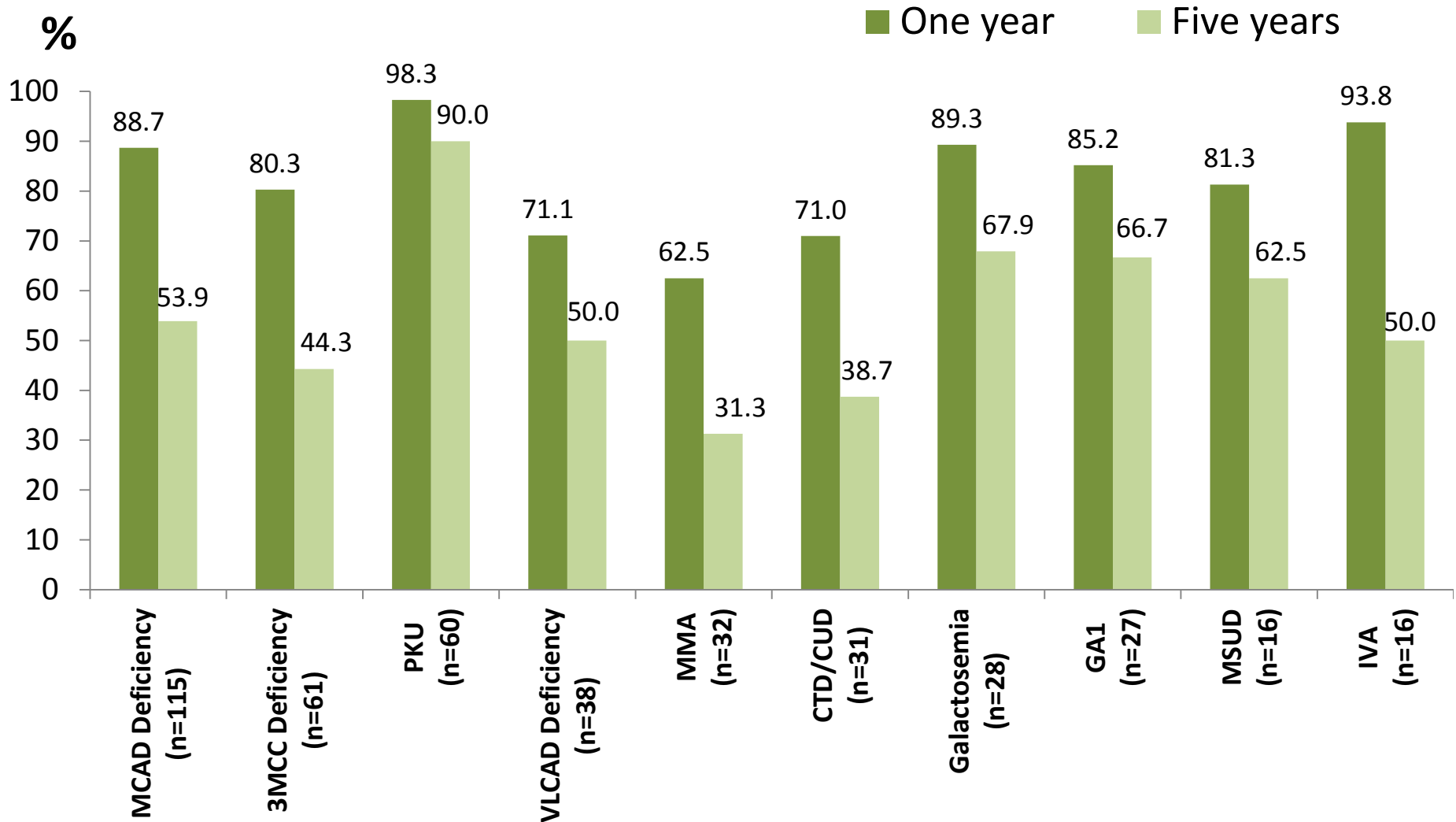
Percent of children remaining in active care in the following year



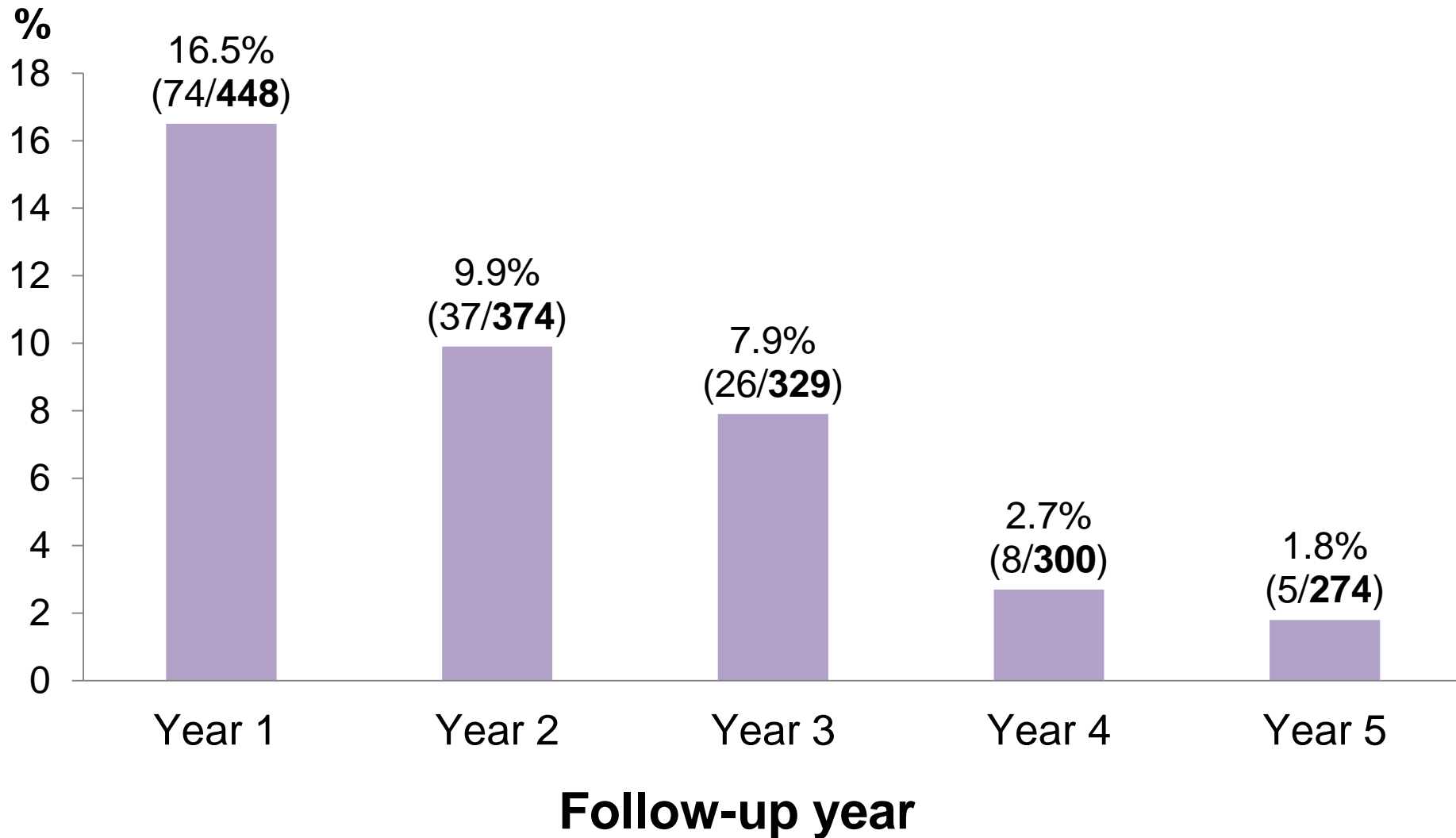
Reported reasons for discontinuation of care by follow-up year



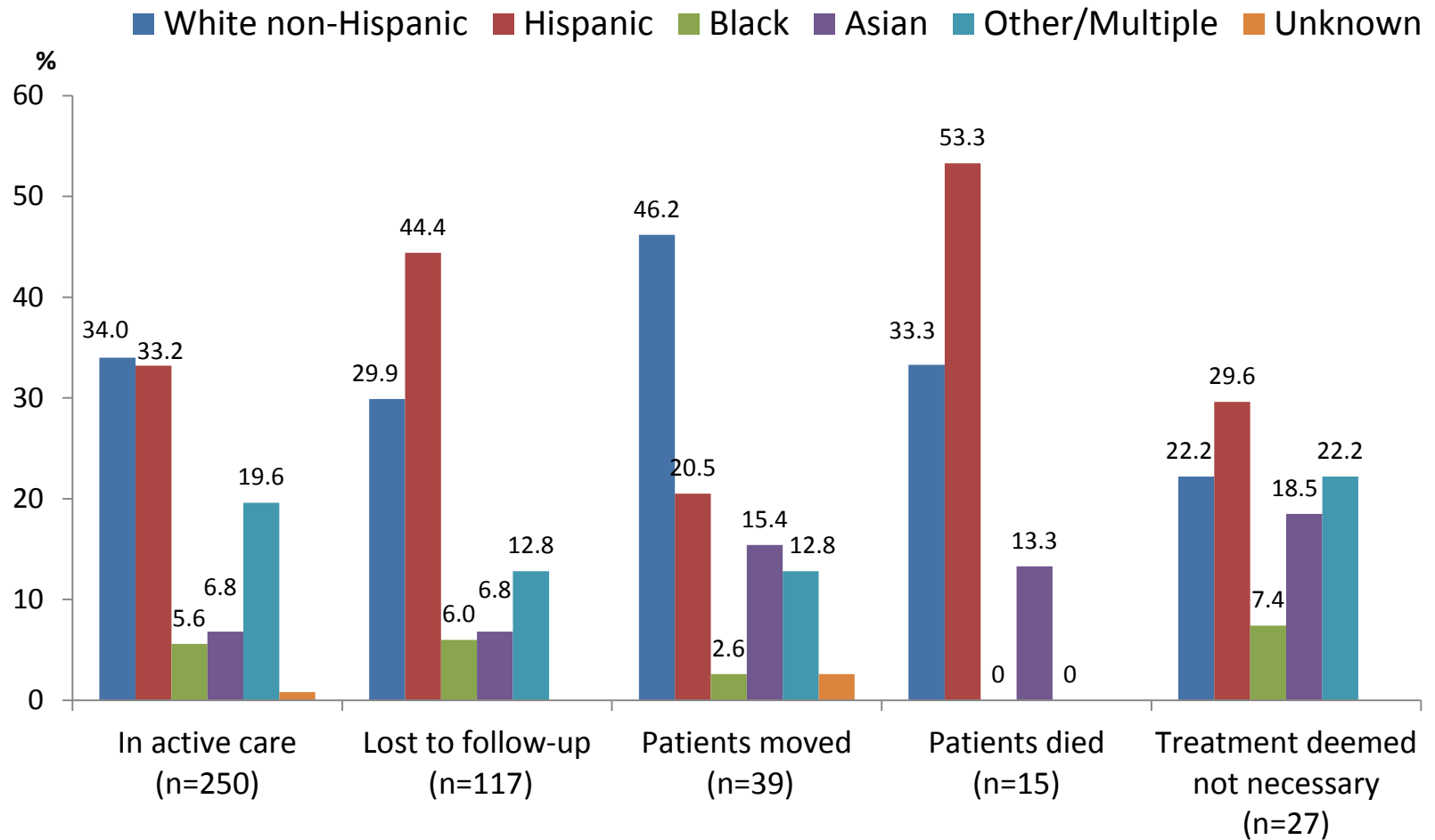
Comparison of one-year and five-year active follow-up status by select disorder



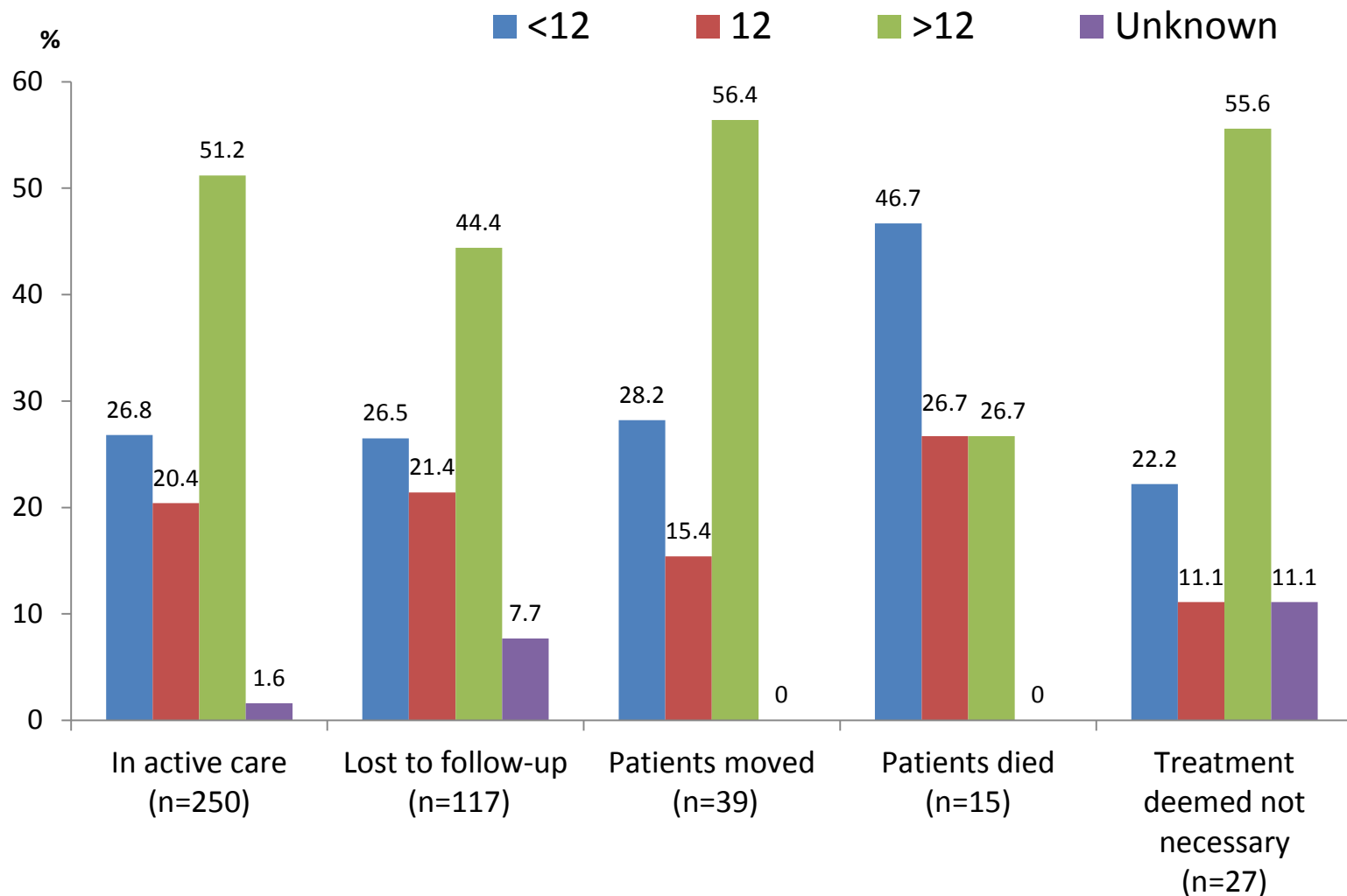
Percentage of missed APS reports among active patients in the following year



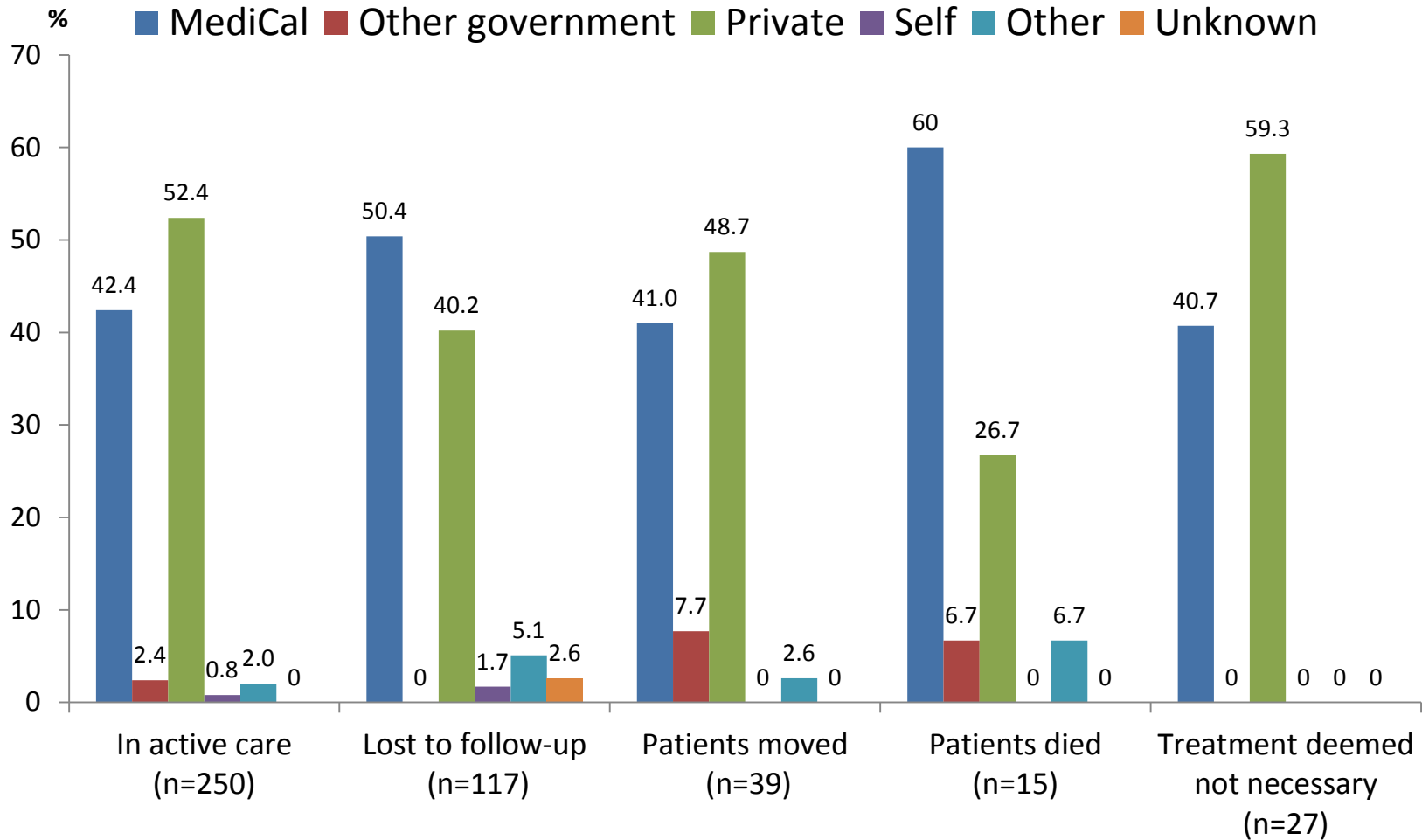
Maternal race-ethnicity for initial cohort and follow-up status



Maternal education (in years) for initial cohort and follow-up status



Health care coverage for delivery for initial cohort and follow-up status



Summary: Active Care

- Of 448 newborns, 56% were in active care by the end of the fifth year of life.
- Certain disorders were associated with a higher percent staying in active care through age five: 90% of PKU, 68% for Galactosemia, 67% for GA1, and 63% for MSUD.
- Disorders associated with lower percent of active care status through age five: 44% for 3MCC, 39% for CTD/CUD and 31% for MMA.
- Most children (70%) who transferred care to another center stayed in active care in the following year.

Summary: Lost to Follow-Up

- More cases were lost to follow-up in the first year of life (8%) compared to subsequent years (5%-7%).
- Over the course of five years, 26% of children were lost to follow-up and 3.3% died.
- Out of a total of 15 deaths, 11 (73%) occurred in the first year of life.
- Two thirds (67%) of lost to follow-ups did not report any health problem in the APS of previous year.

Summary: Maternal Characteristics

- Compared to the Active Care group:
 - Hispanics were more likely to be Lost to Follow-Up.
 - Mothers with more than 12 years of education were less likely to be Lost to Follow-Up.
 - Mothers covered by private insurance were less likely to be Lost to Follow-Up.



Next Steps

- Further exploration of patients that became lost to follow-up
 - Distance to clinic (GIS mapping)
- Detailed analysis by specific disorders
 - Symptoms and developmental status
 - Treatments & services provided
- Affordable Care Act impact on service utilization



Conclusion

- Limitations:
 - Missing data
 - Doesn't capture highly detailed clinical information
- Challenges:
 - Cost of data collection
 - Late-onset disorders
 - Data capture from multiple specialty care centers



Acknowledgement

- Special thanks to the team members at our California state-contracted metabolic centers who provide long term follow-up data for our newborn screening program!

