California Newborn Screening Program

Long-Term Follow Up Data Collection

Lisa Feuchtbaum, DrPH, MPH
Chief, Program Development & Evaluation Branch
Genetic Disease Screening Program
California Department of Public Health

Long-Term Follow Up for Newborn Screening

What is it?

- Systematic evaluation to determine if newborn screening is meeting its goal

Why do it?

- Assurance that condition-specific treatment & age-appropriate preventive care is available for individuals identified with a condition included in newborn screening *

*Kemper, et al. Long–term follow up after diagnosis resulting from newborn screening: statement of the US Secretary of Health and Human Services' Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children. (Editorial) *Genet Med* 2008:10:259-261

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Development of Long-Term Follow Up System

- 2002: A framework for LTFU was created as part of the HRSA funded pilot study to examine the efficacy of MS/MS screening
- 2005: Implementation of a LTFU data system developed as part of a web-based Screening Information System (SIS)
- SIS supports all aspects of lab results reporting, mailer creation, patient referral tracking & coordination with specialty care follow-up centers

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Follow Up Model

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

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

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No Disorder

Confirmed Disorder

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Initiation of Long Term Follow-Up via Annual Patient Summary Data Collection

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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
- State pays for submission of APS reports using SIS
- Reports document whether child is still in active care
- Clinical management strategies
- Clinical outcomes

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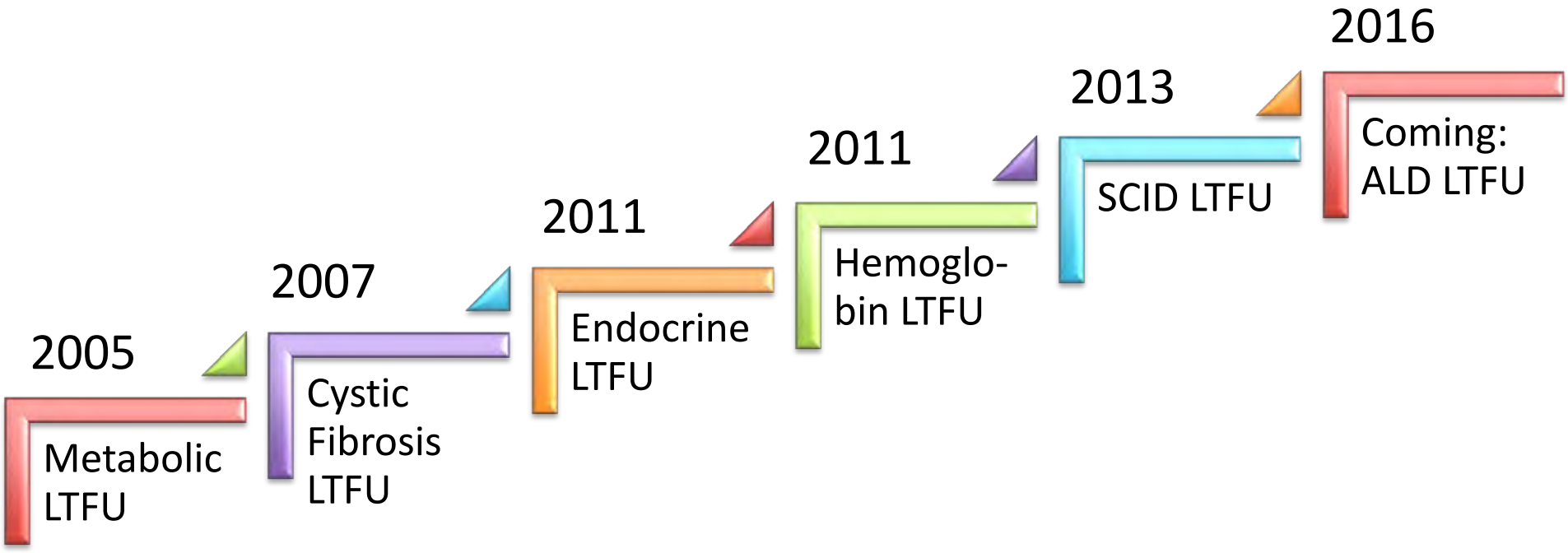
Implementation of Long-Term Follow Up System Since 2005



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10 Years of LTFU Data on Metabolic Disorders*

- Newborns Screened: 5,182,386
- Diagnosed Cases: 1,505
- Total Annual Patient Summaries: 5,208

* As of October 2015

10-Year Count of Annual Patient Summary Reports for Metabolic Disorders

	Age of Child (Years)					
	1	2	3	4	5	All
Disorder Name						
3-Methylcrotonyl-CoA Carboxylase Deficiency (3MCC Deficiency)	109	94	66	47	36	352
Biotinidase Deficiency Partial (BD)	63	57	37	27	19	203
Biotinidase Deficiency Profound (BD)	44	32	29	22	18	145
Carnitine Transporter Deficiency (CTD)/Carnitine Uptake Defect (CUD)	63	52	41	29	19	204
Duarte Galactosemia (D/G)	109	108	54	46	37	354
Galactosemia, classical	26	25	29	23	25	128
Glutaric Acidemia type I (GA1)	45	40	35	28	23	171
Hyperphenylalaninemia, benign	68	60	48	43	33	252
Hyperphenylalaninemia, variant	64	60	55	54	46	279
Isovaleric Acidemia (IVA)	35	26	23	20	15	119
Long Chain Hydroxy Acyl-CoA Dehydrogenase Deficiency (LCHAD deficiency)	8	8	6	7	5	34
Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCAD Deficiency)	182	173	136	112	86	689
Methylmalonic Acidemia mut 0 (MMA)	18	20	18	14	10	80
Methylmalonic Acidemia mut- (MMA)	22	19	11	8	6	66
Methylmalonic Acidemia, Cbl C, D, F (MMA)	36	36	33	31	28	164
Phenylketonuria (PKU)	140	123	112	113	112	600
Short Chain Acyl-CoA Dehydrogenase Deficiency (SCAD Deficiency)	150	122	88	61	44	465
Very Long Chain Acyl-CoA Dehydrogenase Deficiency (VLCAD Deficiency)	52	45	36	29	28	190
All	**1463**	**1277**	**986**	**814**	**668**	**5208**

How Has Long-Term Follow Up Data Been Used?

Collaborative Studies:

- MS/MS Study, Hinton C et al, 4-State Collaborative Study (CDC)
- VLCADD Study, Merritt JL et al. (WSRGC)
- SCADD Study, Galant N, et al. (UCLA)
- 3MCC Study, Lam C, et al. (UCLA)
- CF genotype-phenotype studies, Salinas D, et al (CHLA)
- MS/MS, NIH-funded U19 Study (UCSF)

Long-Term Follow Up Data Uses

What % of children with diagnosed disorders…

- are in care through age five?
- become lost to follow-up?
- have disorder-related complications?
- died and for what reasons?
- have developmental delay?
- have high rates of ER visits & in-patient hospitalizations?
- have more frequent visits to the specialty care follow-up centers?

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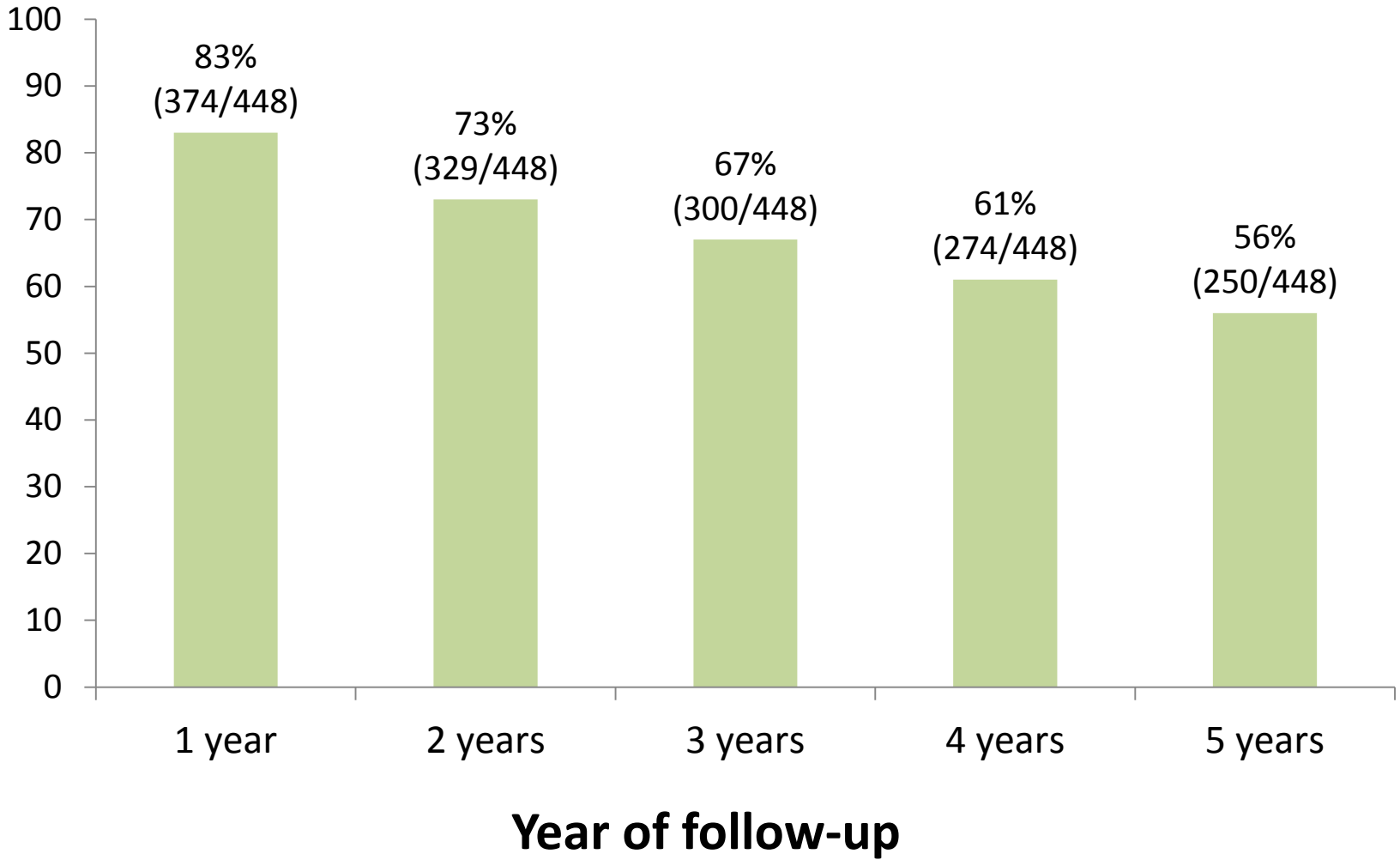
LTFU for Program Evaluation: Access to Care

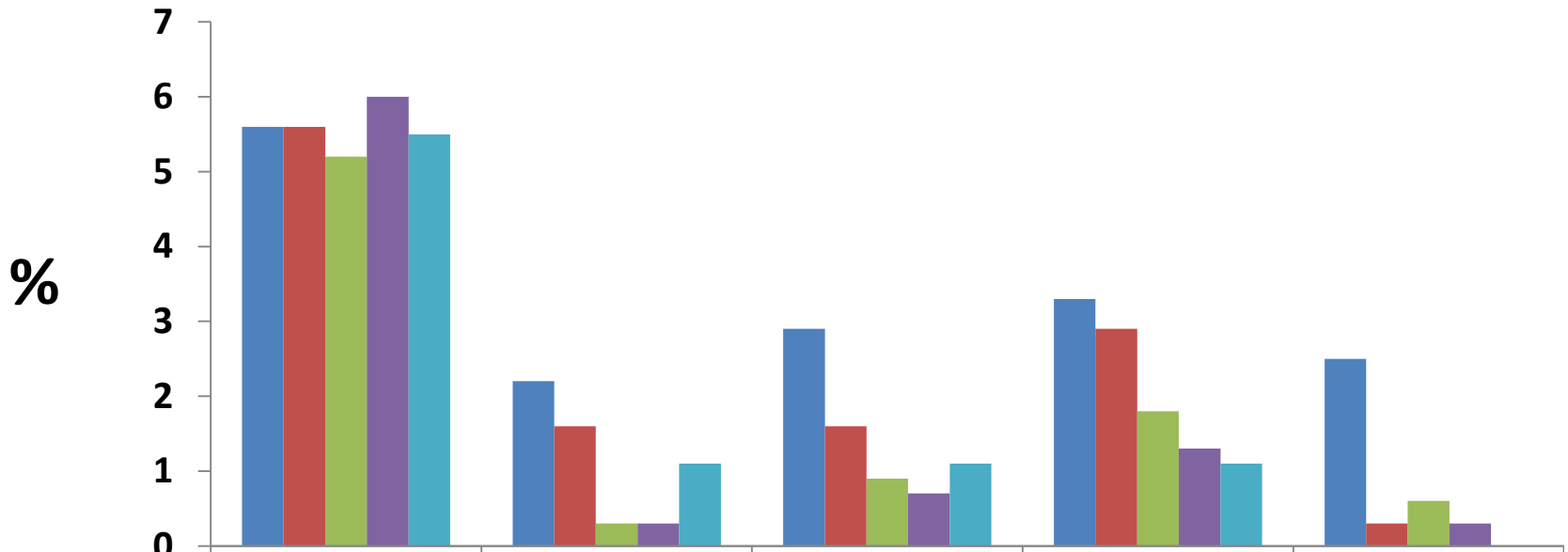
What percent of children with RUSP Primary MSMS disorders remained in active care between the ages of one and five years old?

- 10 years of MS/MS screening: 2 five year cohorts
- 2,514,004 newborns screened between 07/07/2005 to 07/06/2015.
- 448 RUSP Primary Metabolic Disorders diagnosed

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

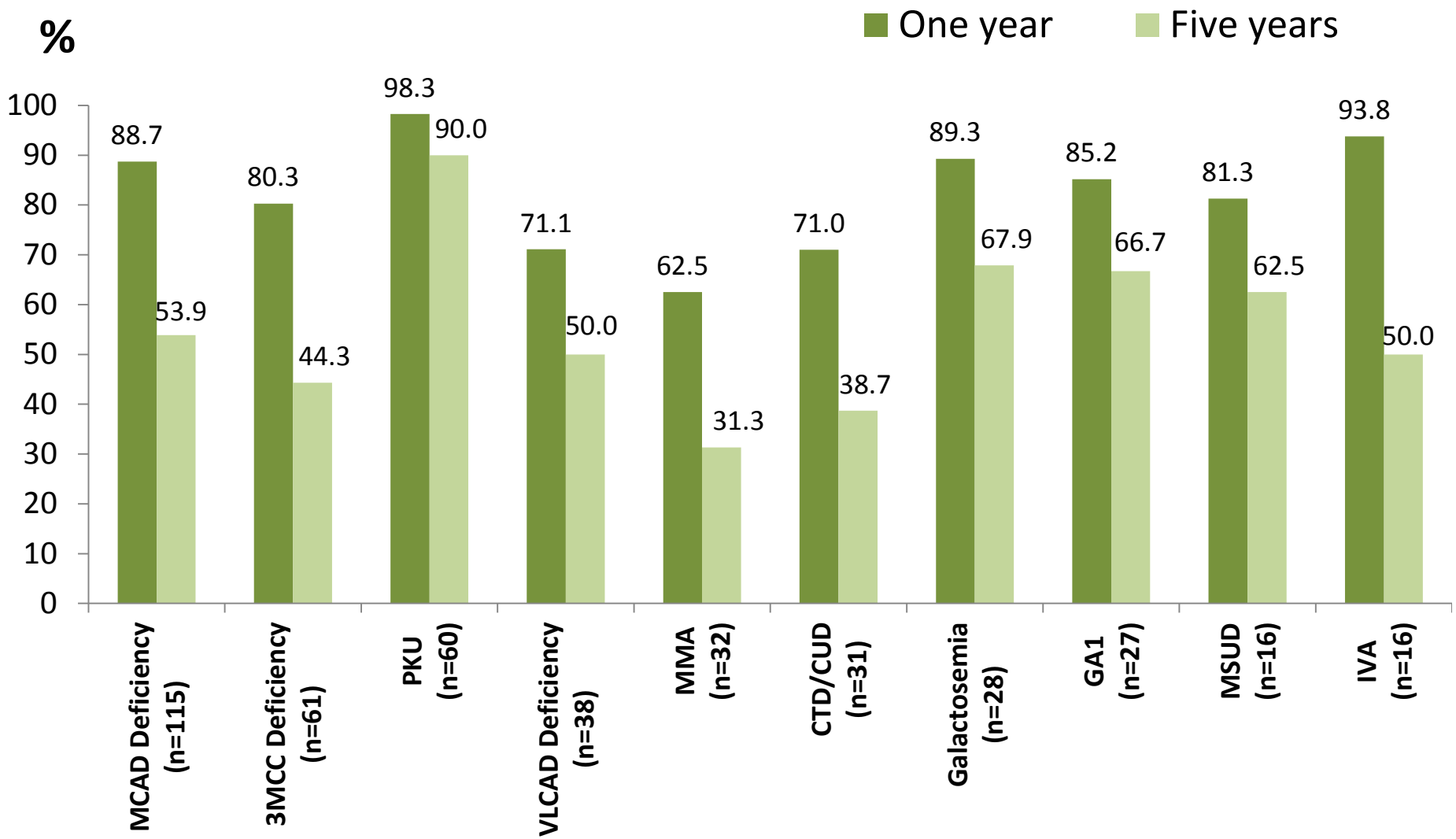


Reported reasons for discontinuation of care by follow-up year

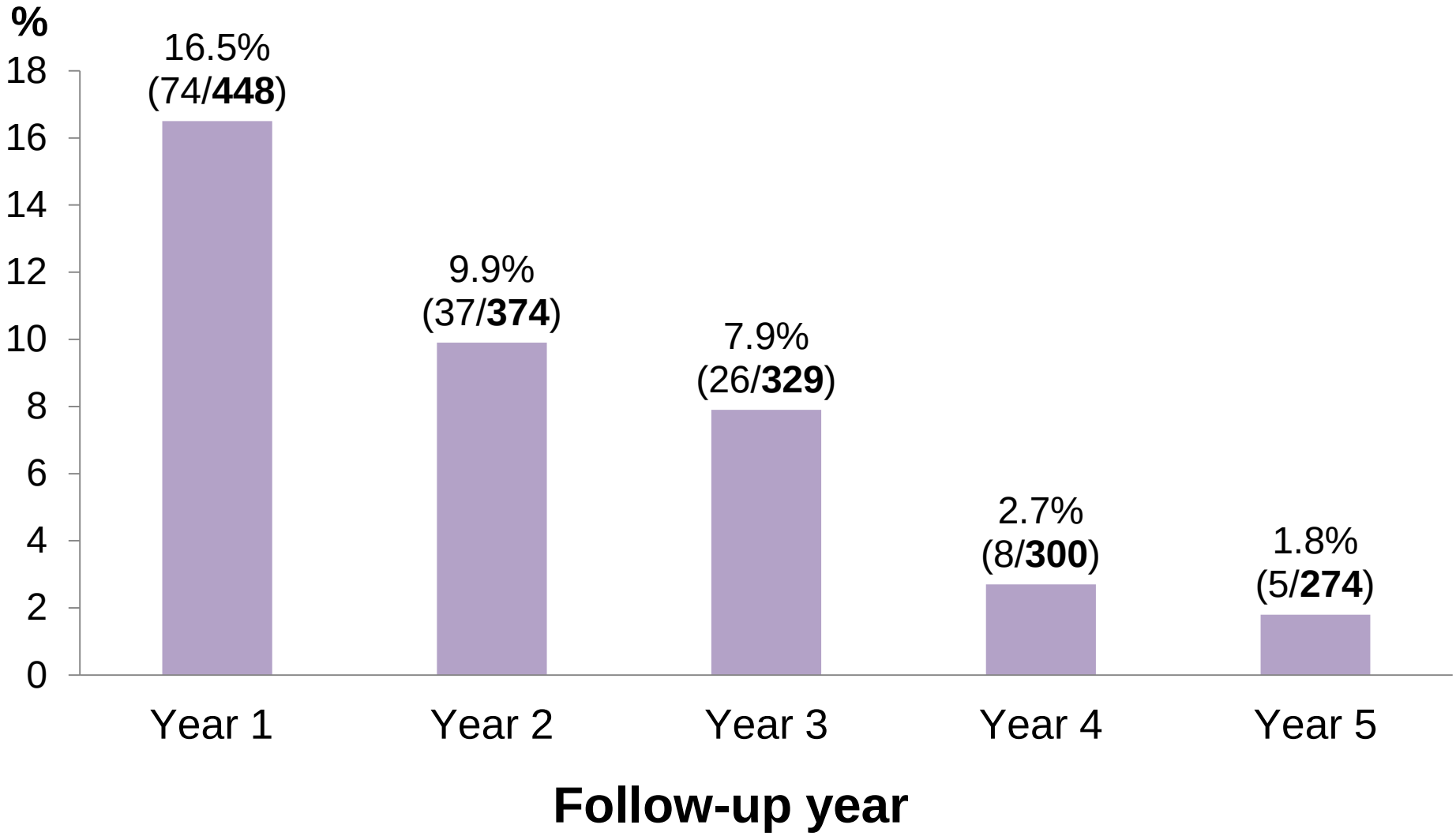


	Lost to follow-up	Refused follow up	Treatment deemed not necessary	Move-out-of state	Child died
Year 1 (n=448)	5.6	2.2	2.9	3.3	2.5
Year 2 (n=374)	5.6	1.6	1.6	2.9	0.3
Year 3 (n=329)	5.2	0.3	0.9	1.8	0.6
Year 4 (n=300)	6.0	0.3	0.7	1.3	0.3
Year 5 (n=274)	5.5	1.1	1.1	1.1	0

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



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

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Conclusion

- LTFU provides data on impact of newborn screening programs
- A valuable resource for clinical collaborations and program evaluation
- 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

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