# Long-term Follow-up for Cystic Fibrosis: A Collaboration with the Cystic Fibrosis Foundation



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# Outline

- Introduction
- Matching
- Long-term follow-up

# Introduction

○ Long-term follow-up data already entered into the Cystic Fibrosis Foundation Patient Registry

  ● Consented Registry

○ Plan: request data from CF Foundation on patients born in NYS

# Introduction

- What long-term follow-up questions are important from a public health standpoint?

# Introduction

○ Question 1: Are there individuals with CF in NYS who had a false negative newborn screen and when were they diagnosed?

○ Question 2: What percentage of children in NYS enter and remain in specialty care after receiving a CF diagnosis?

○ Question 3: How many children with cystic fibrosis move into and out of NY?
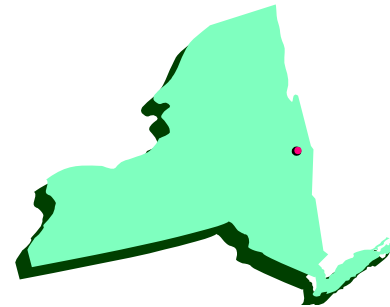
# Introduction

- Data requested
  - State of birth
  - Patient's date of birth
  - Patient Registry ID
  - All fields in the CF diagnosis category
  - Data year
  - All encounter fields
  - Died in data year

# Introduction

- Data years
  - Patients born from first complete year of CF screening in NYS (2003) to 2010
  - Submit additional materials to receive data in the future

# Question 1

- Are there individuals with CF in NYS who had a false negative newborn screen and when were they diagnosed?

# Methodology – Matching

- Match NBS data to CFFPR data
- Goal: Identify false negatives and evaluate matching criteria
- Match criteria: date of sweat test, sweat chloride value, date of birth, gender and diagnosis

# Results

- Received data for 369 cases from 2003 to 2010
- Number of cases in NBS data: 351

# Results

- 68/369 Cases that did not match in NBS data and CFFPR data
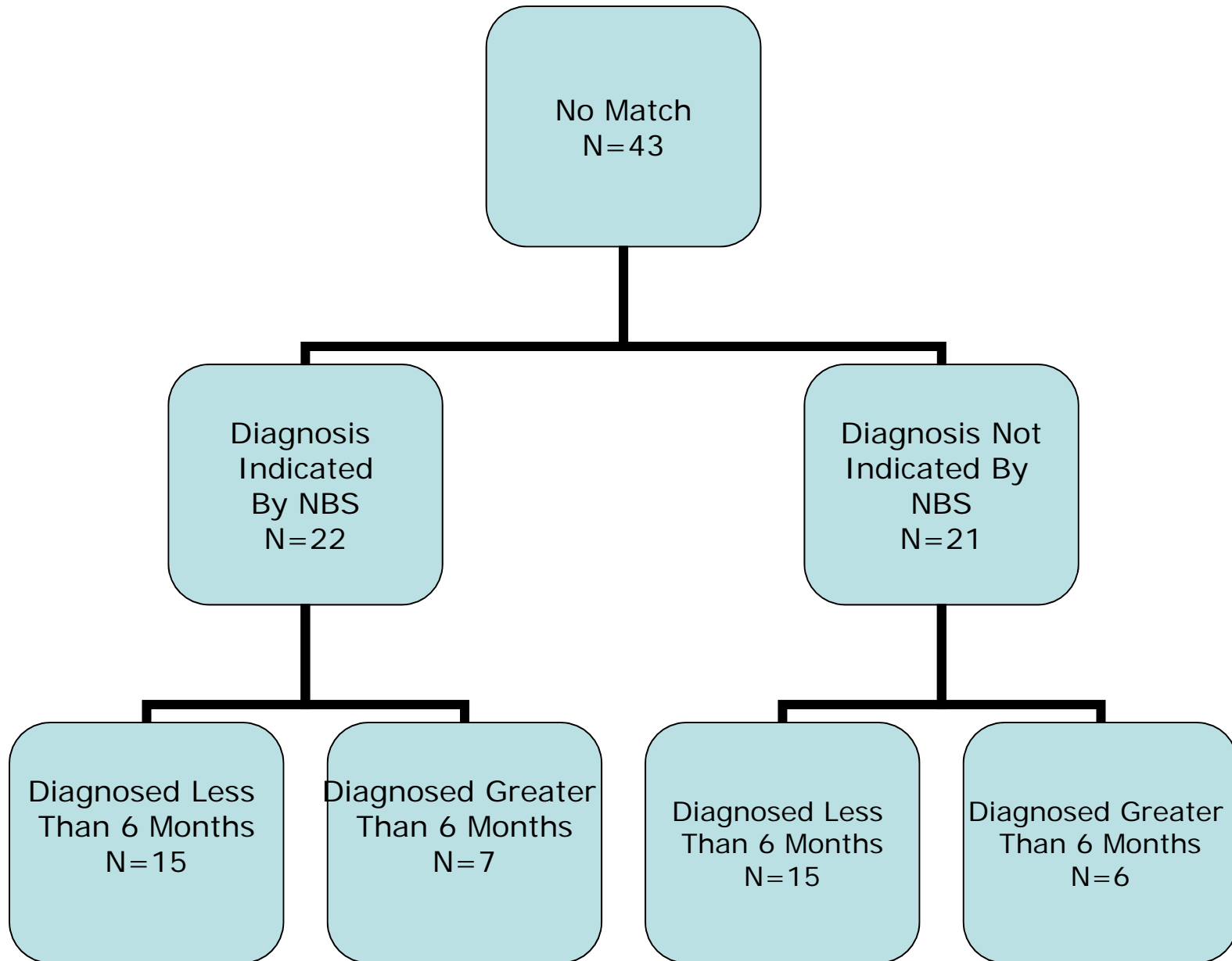- 81.6% match rate
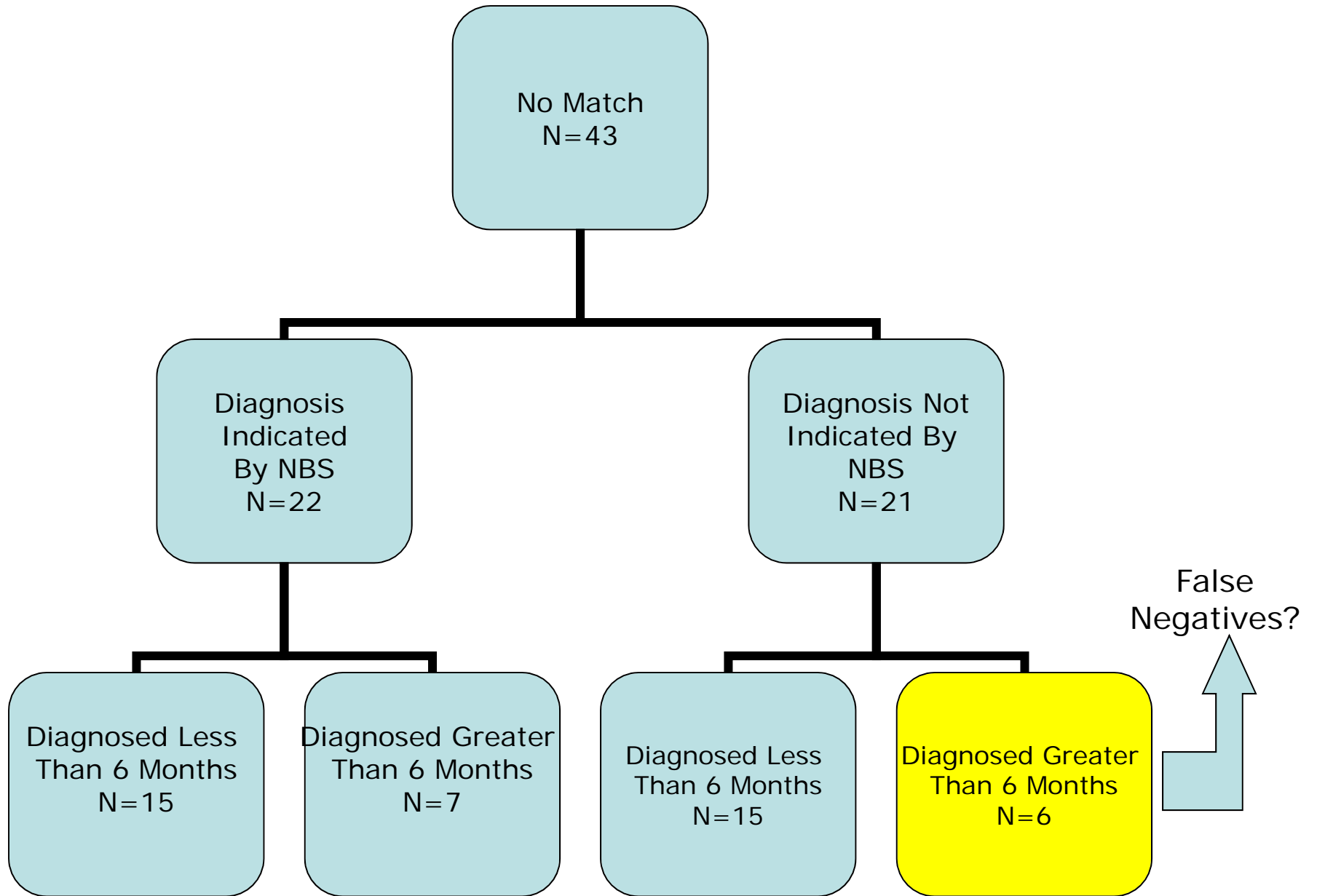
# Results

- 25 of 68 cases had a likely match
  - Considered likely match if:
    - Matching DOB and gender
    - Different sweat test data or diagnosis
    - Average age of diagnosis 0.29 years (0.044-1.47)

# Results

- No match
  - 43 cases
  - Average age of diagnosis 0.85 years
  - Considered no match if:
  - No newborn in NBS record with a CF diagnosis and matching DOB and gender

# Results

- Diagnosis greater than 6 months and not indicated by newborn screening
  - N=6
  - Most likely false negatives
  - Average age of diagnosis – 3.24 years
  - One case previously known to NBS as false negative

# Discussion -

○ Further investigation into these 6 cases

- Root cause analysis
- Is there a reasonable corrective action?
  - ○ Consider: impact on false positive rate

# Discussion

- No match: diagnosed <6 months or diagnosis indicated by NBS
  - N=37
- Likely represent limitations of matching

# Discussion: Limitations of Matching

- Data quality
  - Data entry errors
- Case closed as lost to follow-up
- NBS receiving diagnosis prior to genetic test results available
- Case closed based on sweat value only
- Case closed as expired without diagnosis
- Positive NBS in another state
- Not screened

# Discussion

- Source of Bias
  - False negatives that have not been diagnosed yet or have not been entered into the CFFPR

# Conclusions

- Unique identifier would help with analysis and interpretation
  - Sweat test not always good for matching because it is sometimes repeated after diagnosis form sent to NBS

# Questions

- Question 2: What percentage of children in NYS enter and remain in specialty care after receiving a CF diagnosis

# Methods - Long-term Follow-up

- Data from CFFPR: Year and number of encounters for each patient
- Encounter after the diagnosis date?
- Encounter in 2010?
- Year of life the last encounter occurred?

# Results - Long-term Follow-up

- Confirmed visit at CF Center after diagnosis
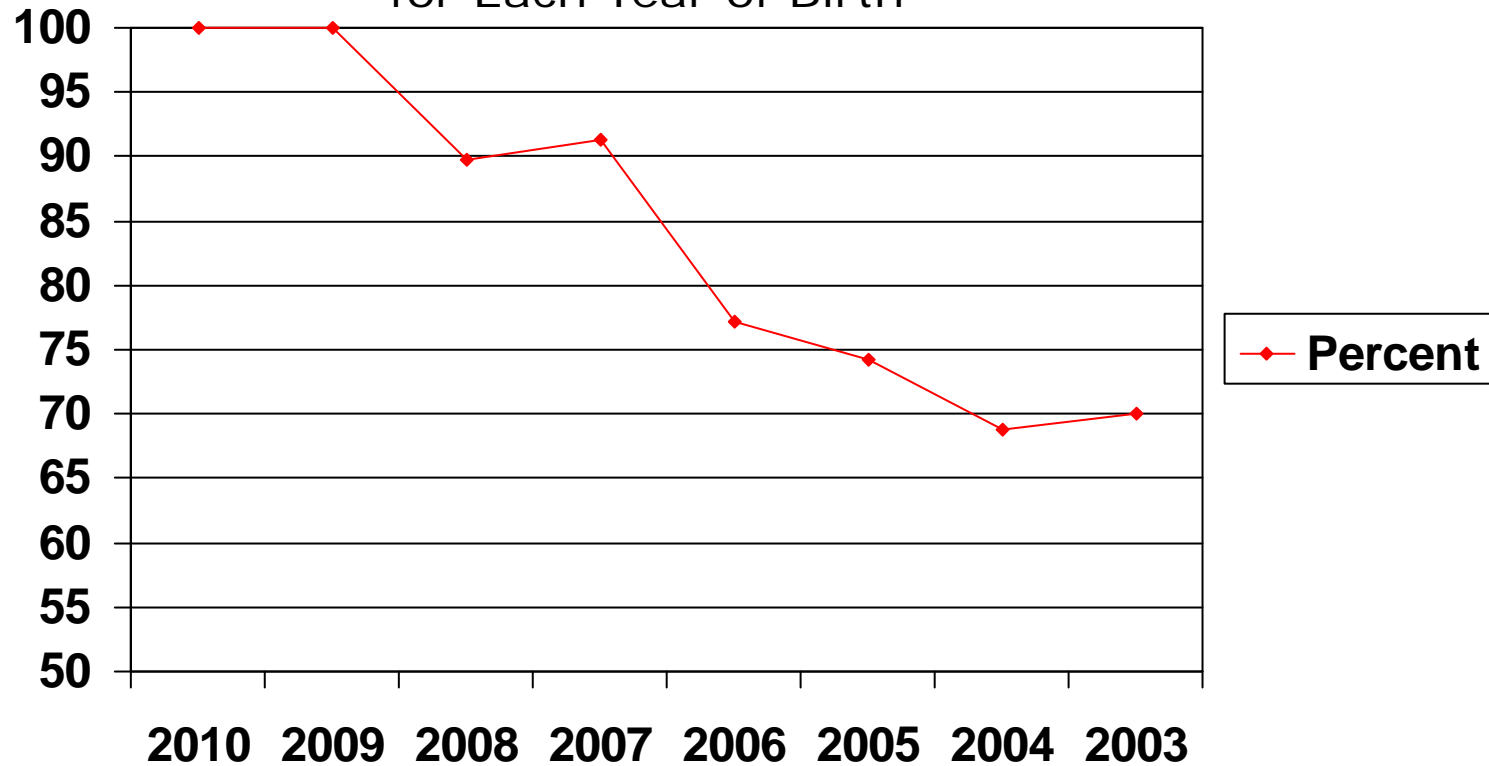  - 357 out of 368 (97%)

# Results - Long-term Follow-up

- 63 out of 366 (17%) did not have a visit in the CFFPR in 2010
  - possibly lost to follow-up, discharged from care or not updated in registry
  - Deceased patients were excluded (N=3)

# Long-term Follow-up Results

Percent Patients with an Encounter in CFFPR in 2010
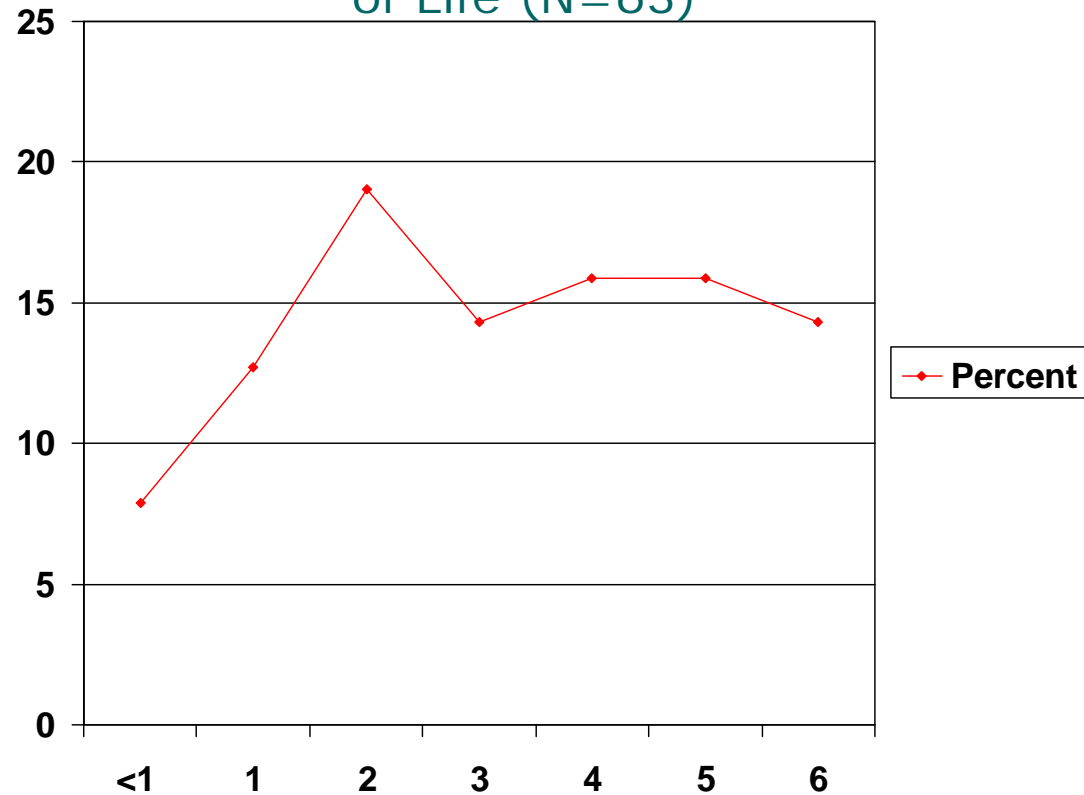for Each Year of Birth

# Long-term Follow-up - Results

| Year of Life | Number of Patients with Their Last Encounter in the Year | Percent of Total (N=63) |
|---|---|---|
| <1 | 5 | 7.9 |
| 1 | 8 | 12.7 |
| 2 | 12 | 19.05 |
| 3 | 9 | 14.29 |
| 4 | 10 | 15.87 |
| 5 | 10 | 15.87 |
| 6 | 9 | 14.29 |

# Long-term Follow-up - Results



Percent of Patients Whose Last Encounter in the CFFPR Occurred During Each Year of Life (N=63)

# Discussion – Long-term Follow-up

- At least 90% of patients born in 2007 and later had a visit in the CFFPR in 2010

- More studies needed to determine why ~17% of patients did not have a visit in 2010

# Question

- Question 3: How many children with cystic fibrosis move into and out of NY?

# Results – Long-term Follow-up

- Patients that moved out of state
- 15 patients moved out of state
  - 15/369 – 4.1%
  - Oregon, Ohio, Texas, South Carolina, Florida, Michigan, Connecticut (3), Kentucky, New Jersey, New Hampshire, North Carolina, Utah, Vermont

# Results – Long-term Follow-up

○ Live in Another State

○ 15/369 – 4.1%

- New Jersey (6), Colorado, North Carolina (3), Pennsylvania, Virginia, Connecticut, Maryland, Florida

# Results – Long-term Follow-up

- 6 moved into NY from another state
  - All born in NY
  - Vermont (2), New Hampshire (2), Connecticut, Missouri

- 18 with an unknown state of residence for initial entry in CFFPR
  - All born in NY

# Conclusions

○ Data would not be available for approximately 8% of patients if it was not from the CFFPR

- Could be as low as 4.6% if patients in border states are followed at NY Center

# Acknowledgements

- Funding Support through HRSA 09-242
- Cystic Fibrosis Foundation

# Thank you!
# Questions?