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
No Match
N=43

Diagnosis Indicated By NBS
N=22

Diagnosed Less Than 6 Months
N=15

Diagnosed Greater Than 6 Months
N=7

Diagnosis Not Indicated By NBS
N=21

Diagnosed Less Than 6 Months
N=15

Diagnosed Greater Than 6 Months
N=6
No Match
N=43

Diagnosis Indicated By NBS
N=22

- Diagnosed Less Than 6 Months
  N=15

- Diagnosed Greater Than 6 Months
  N=7

Diagnosis Not Indicated By NBS
N=21

- Diagnosed Less Than 6 Months
  N=15

- Diagnosed Greater Than 6 Months
  N=6

False Negatives?
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

<table>
<thead>
<tr>
<th>Year of Life</th>
<th>Number of Patients with Their Last Encounter in the Year</th>
<th>Percent of Total (N=63)</th>
</tr>
</thead>
<tbody>
<tr>
<td>&lt;1</td>
<td>5</td>
<td>7.9</td>
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<tr>
<td>1</td>
<td>8</td>
<td>12.7</td>
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<td>4</td>
<td>10</td>
<td>15.87</td>
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<tr>
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<td>15.87</td>
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<tr>
<td>6</td>
<td>9</td>
<td>14.29</td>
</tr>
</tbody>
</table>
Long-term Follow-up - Results

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

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- Cystic Fibrosis Foundation
Thank you!
Questions?