DATA CHALLENGES

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GOALS TO IMPROVE TIMELINESS

- Decrease the time to:
  - Reporting results
  - Getting confirmatory testing
  - Making a presumptive diagnosis
  - Making final diagnosis

- To measure improvement we need consistent metrics
  - CF Clinical Community
  - Public Health Newborn Screening Community

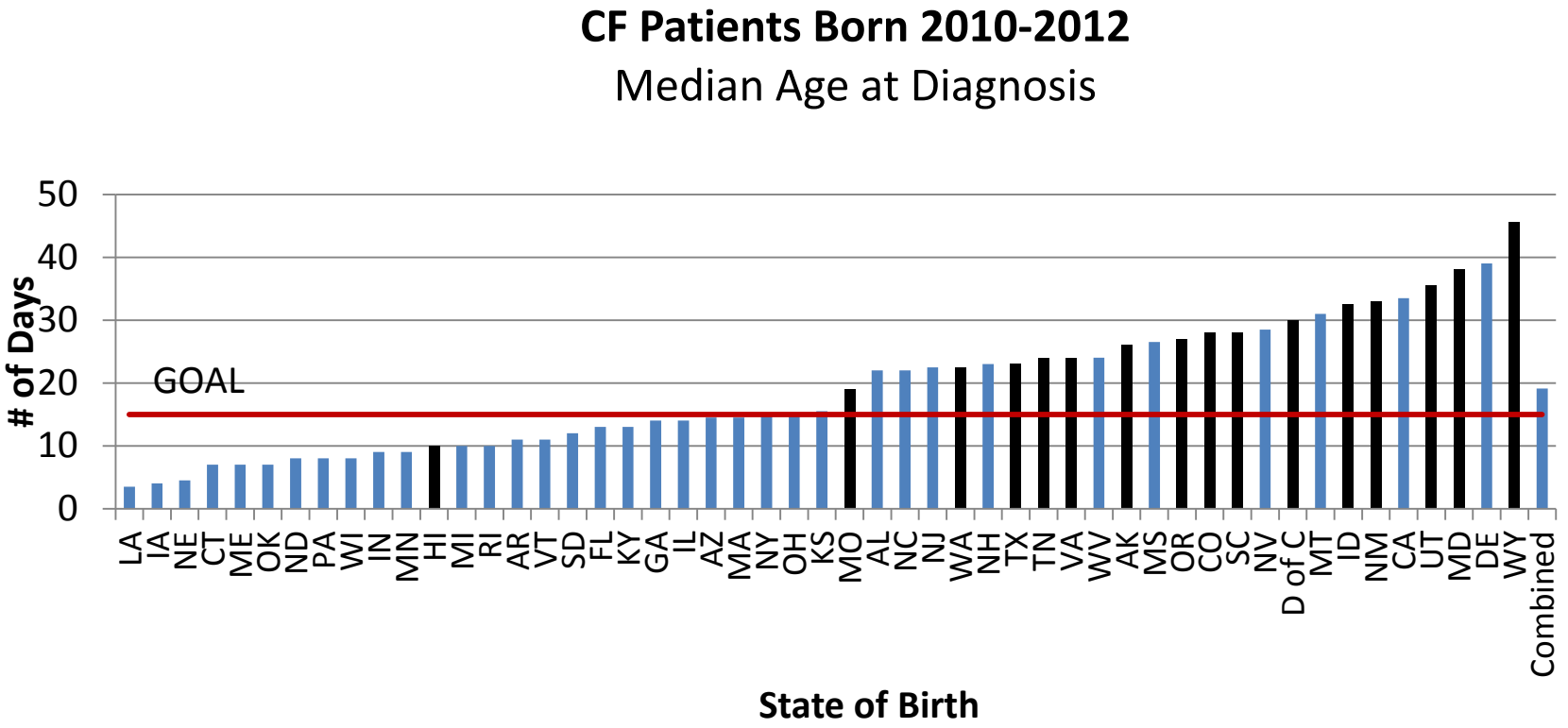
Cystic Fibrosis Foundation Patient Registry Data

CF FOUNDATION PATIENT REGISTRY

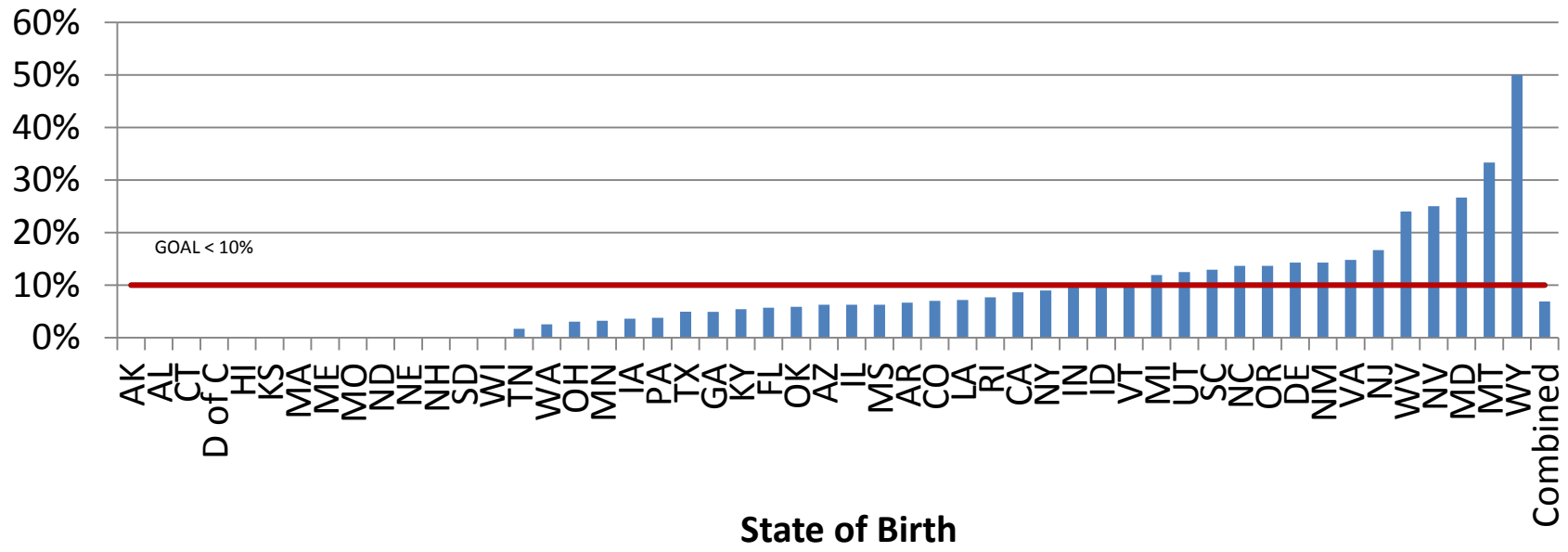
- CF Foundation-accredited care centers
- Health status of people with cystic fibrosis
- Consent based
- Visit- based entry
- Used to create CF care guidelines, assist care teams providing care to individuals with CF and guide quality improvement initiatives at care centers.
- Researchers also use the Patient Registry to study CF treatments and outcomes and to design CF clinical trials.

https://www.cff.org/Our-Research/CF-Patient-Registry/

AGE OF DIAGNOSIS



CF PATIENTS BORN 2010-2012
% DIAGNOSED > 60 DAYS
POSITIVE NBS



NewSTEPs Data

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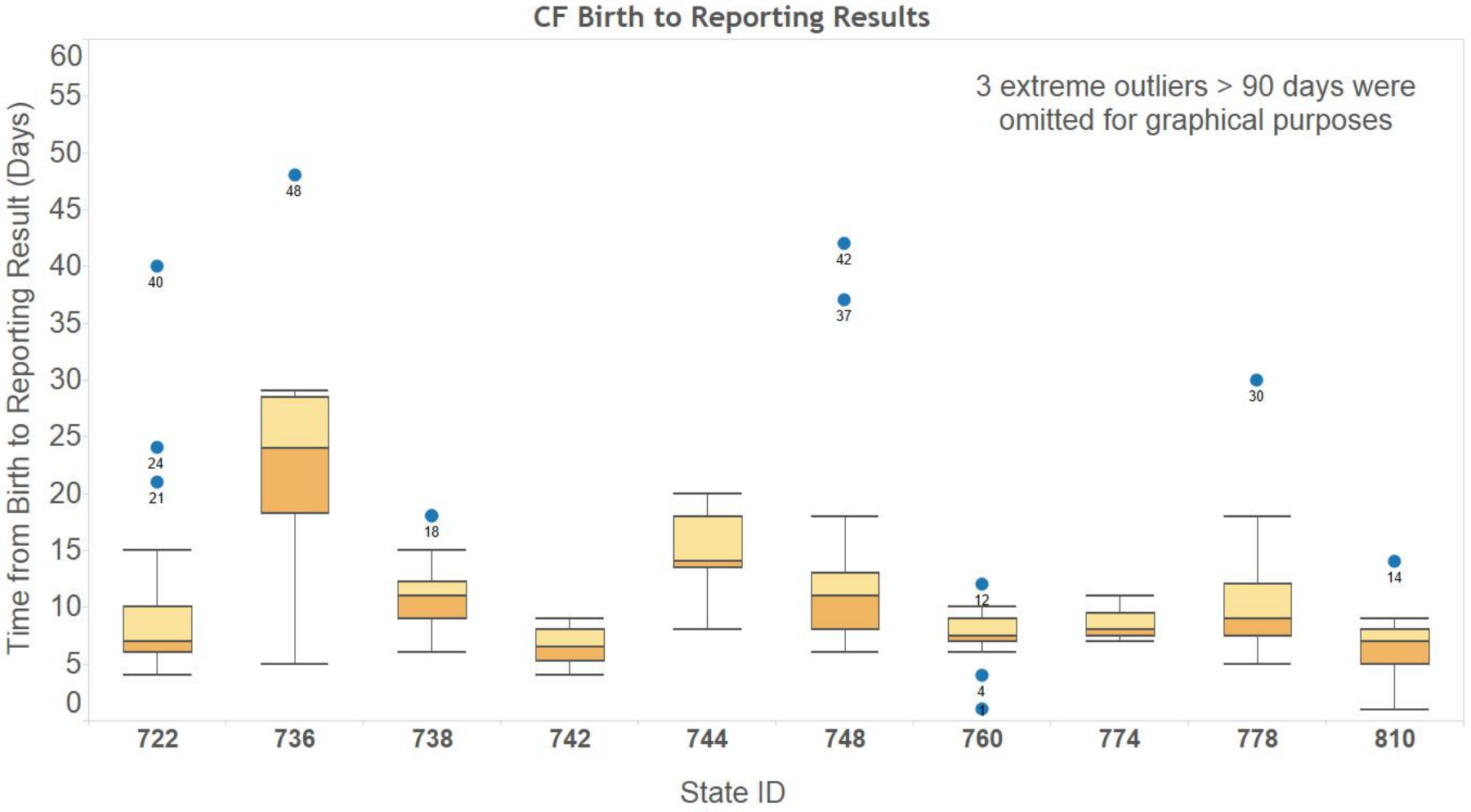
NEWSTEPS DATA REPOSITORY

- State/territorial public health newborn screening programs
- Quality indicators and outcomes of newborn screening programs
- Confidential data reports
- Used to create reports for quality improvement and program management within a state
- Positive newborn screening with diagnosis case level data entry – newborn screening metrics

Cystic Fibrosis
Time to Release of Out of Range Results



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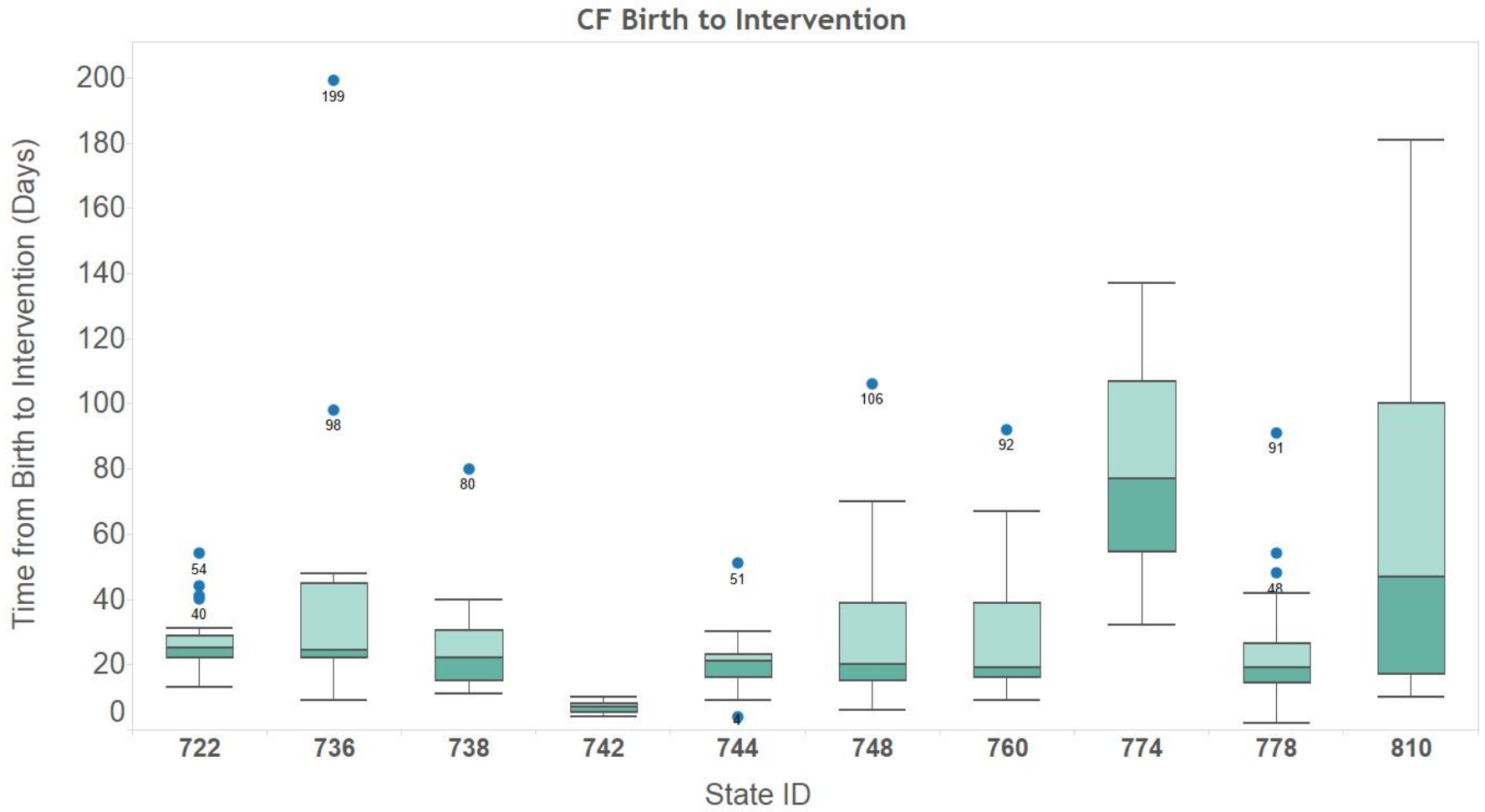
Cystic Fibrosis
Time to Intervention by State



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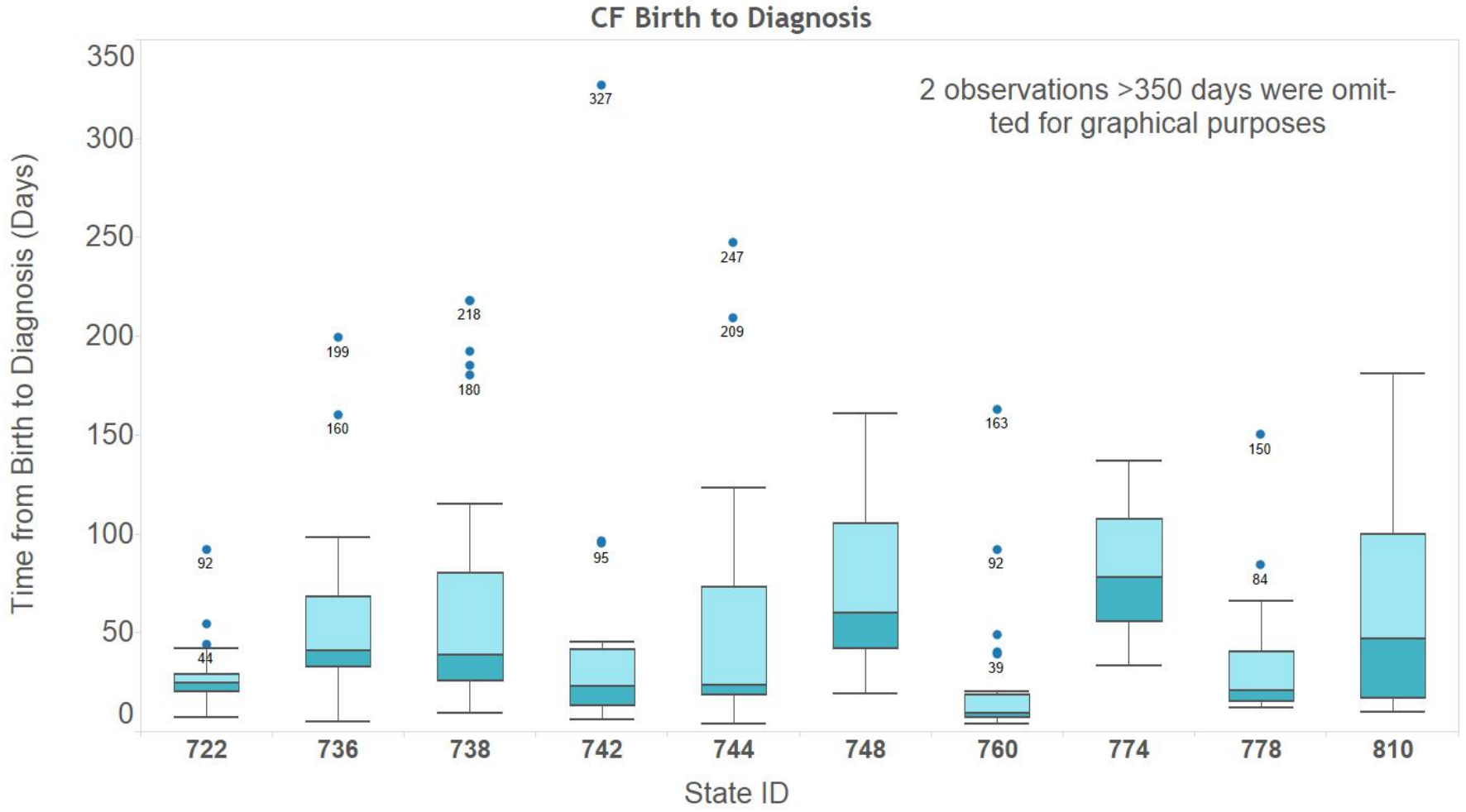
Cystic Fibrosis
Time to Confirmed Diagnosis by State



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DATA DEFINITIONS BETWEEN PROGRAMS NOT CONSISTENT

- CF Centers
  - Diagnosis is defined by Consensus Guidelines, but not consistently implemented by CF Center Clinicians
  - Date of first intervention not utilized historically
- Public Health Newborn Screening Programs
  - Typically depend upon CF Centers for diagnosis date
  - Date of intervention is not well defined

- Date of Intervention is a critical outcome for both programs.

COMMON DEFINITIONS - CF

- Date of intervention (earliest point at which a clinical decision was rendered based on the *presumptive* diagnosis of CF):
  - Therapies may include Enzymes/Salt
  - Documentation of phone conversation that changed the care of that infant
  - Clinic visit
  - This should be inclusive of date therapy was initiated or a decision was made to defer therapy based on current presentation
- Date of diagnosis, with diagnosis confirmed upon:
  - Positive sweat chloride test (Cl $\geq$ 60 mmol/L) to confirm out-of-range screening result
  - Genotype/sequencing to identify CFTR mutations (on sample taken from the infant)
  - Result of Nasal Potential Difference Results

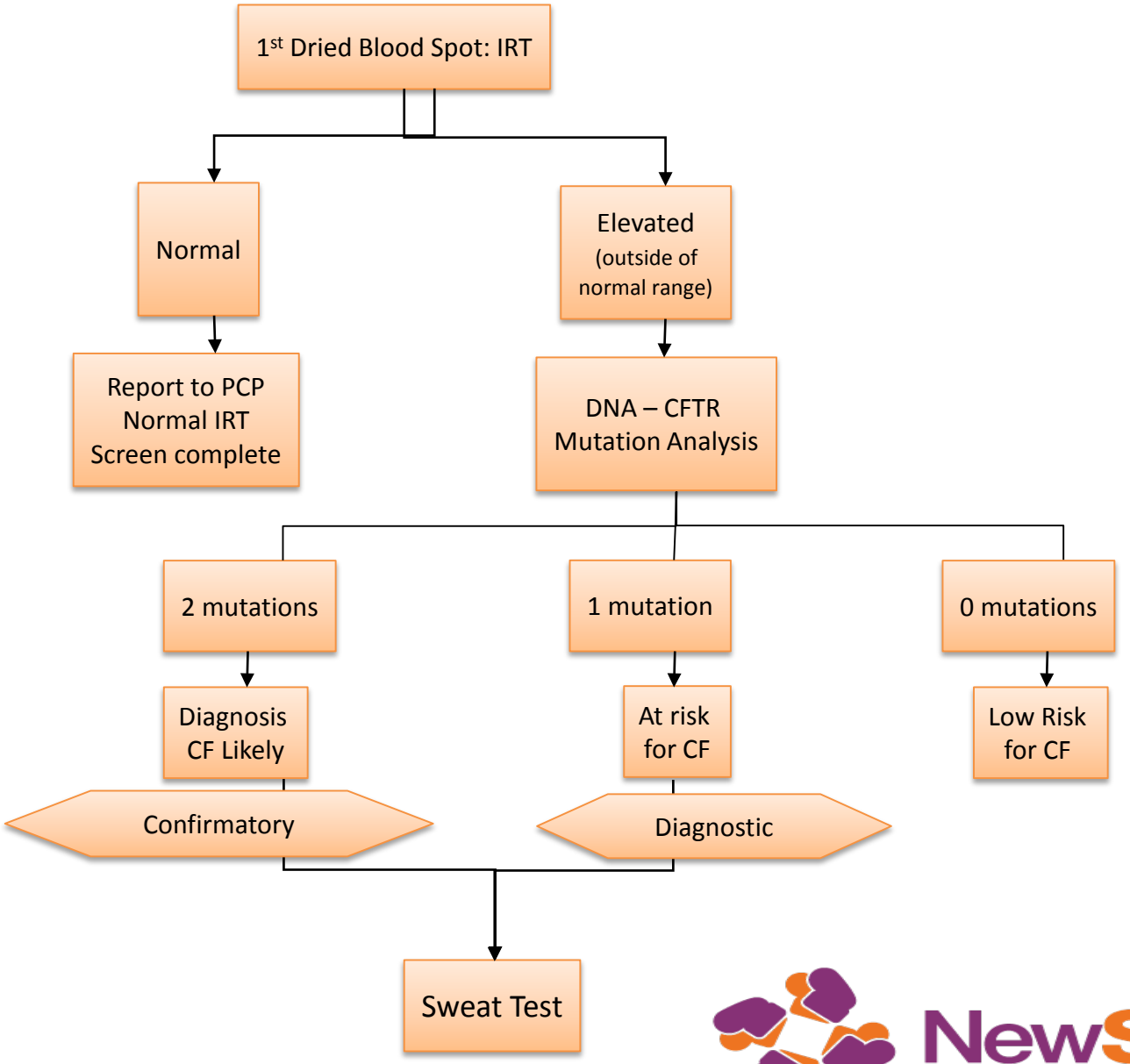
* Presumptive diagnosis of CF following a newborn screen result is one in which the newborn screening results indicate a high likelihood of a diagnosis of CF, but a final diagnosis has not yet been made (for example: elevated IRT and a genotype of phe508del/phe508del)

CFF DIAGNOSTIC GUIDELINES

- Currently under revision and preparation for publication (to occur in fall 2016)

ALGORITHMS

IRT/DNA



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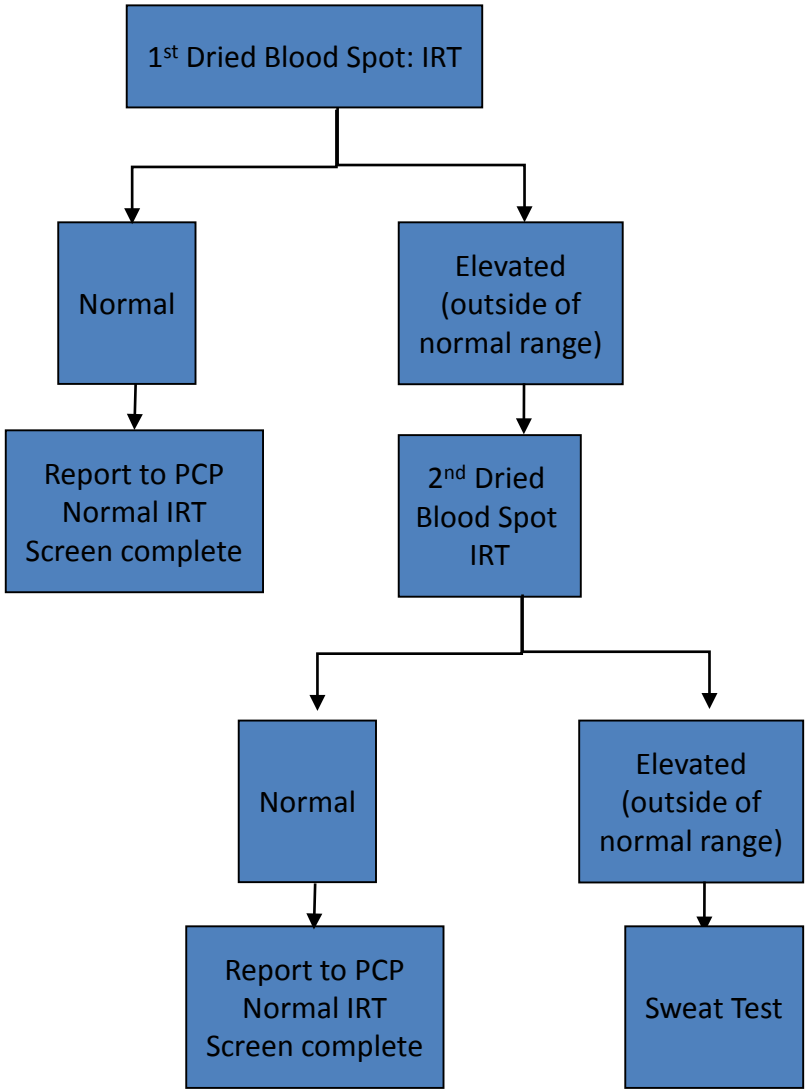
IRT/IRT



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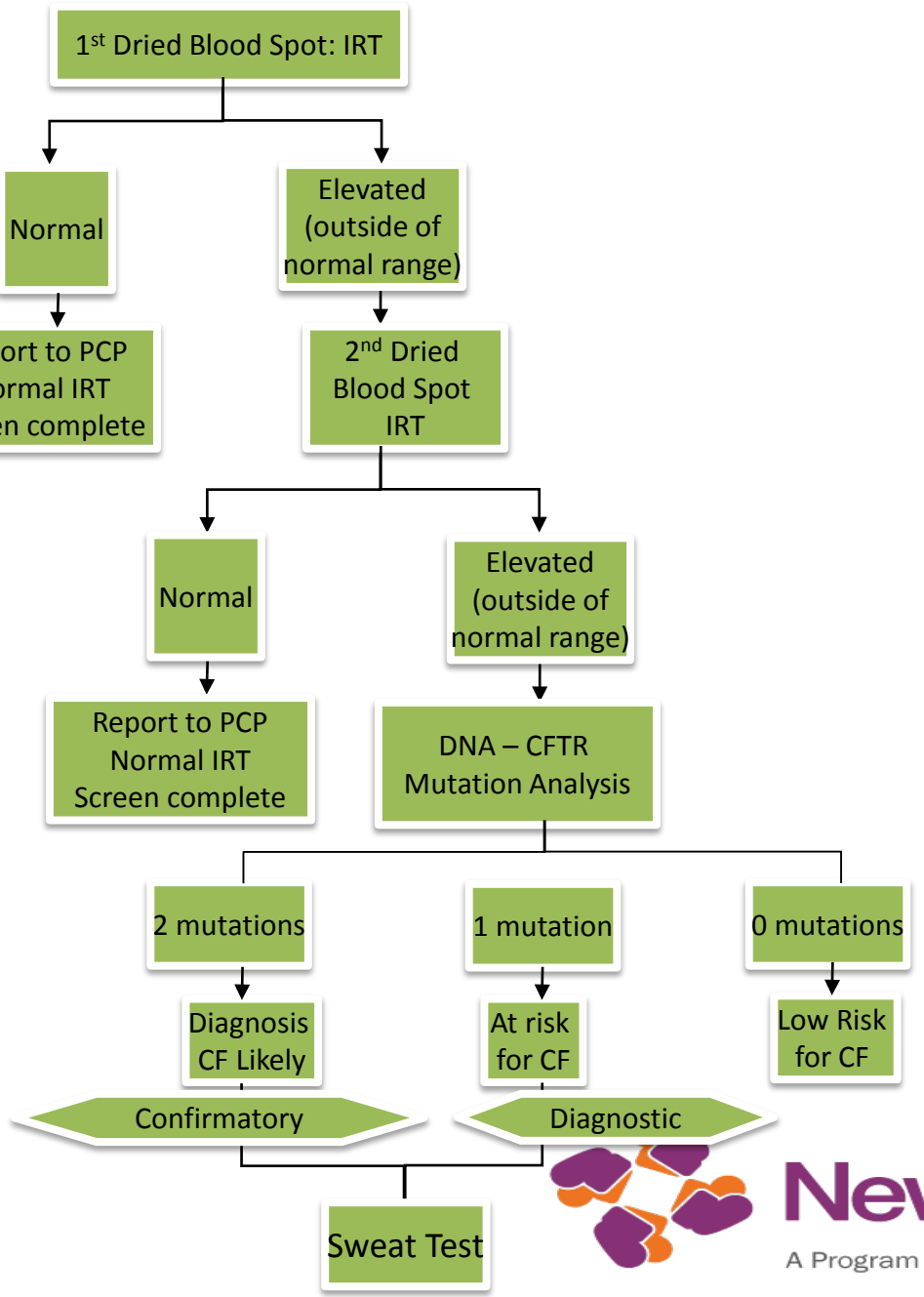
IRT/IRT/DNA



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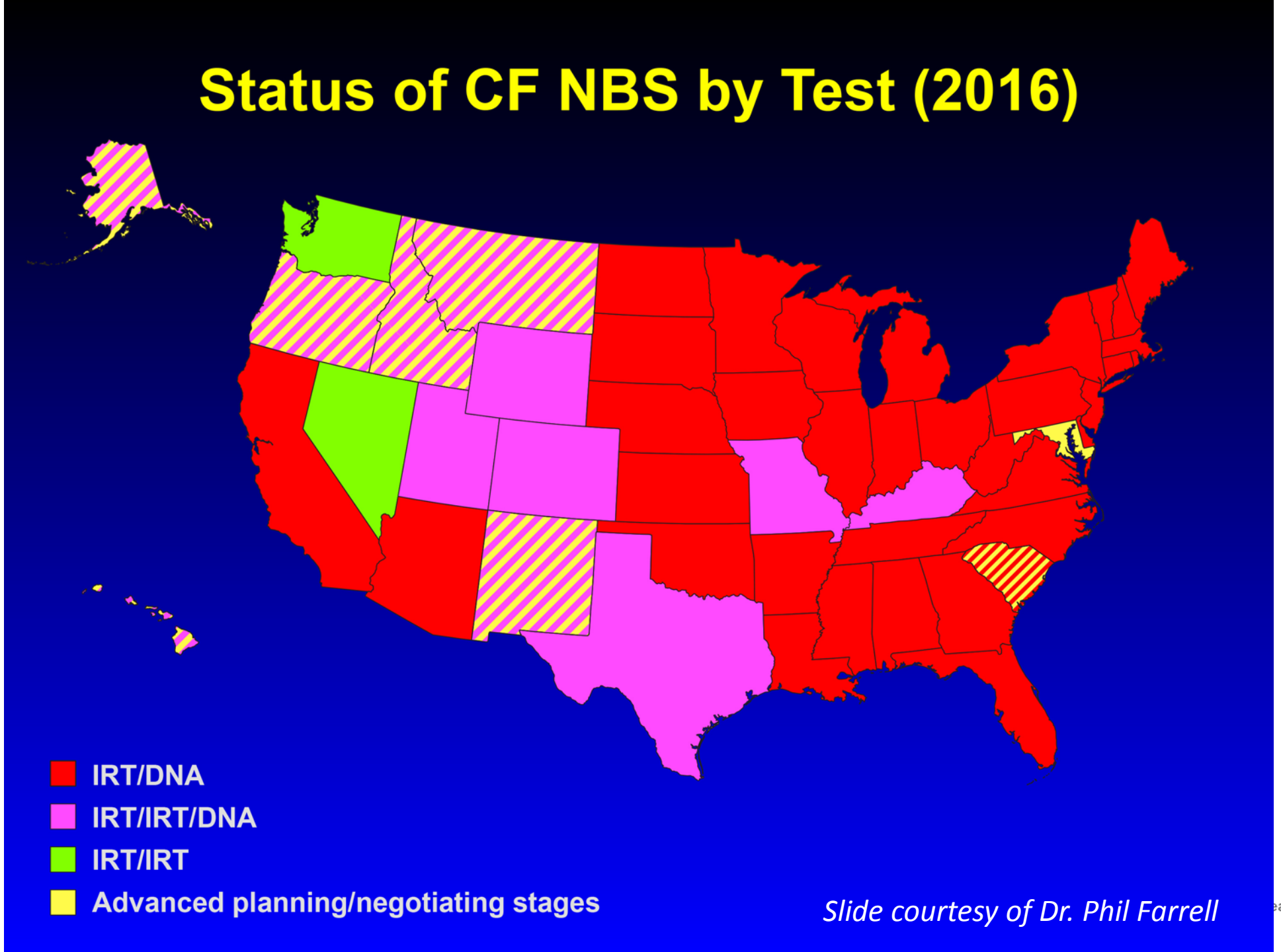
Status of CF NBS by Test (2016)



Slide courtesy of Dr. Phil Farrell

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ALGORITHM DISCUSSION

- TIMELINESS:
  - What challenges are specific to your CF algorithm (IRT/DNA; IRT/IRT or IRT/IRT/DNA) for timely NBS?
  - Who is involved in the screen? Follow-up? Call-out? Diagnostic workup?
  - How does molecular testing as a second tier on the NBS panel impact the timely reporting/diagnosis?
  - What are potential solutions specific to your algorithm?
- Outside the scope of the morning's discussion
  - Discussion of cutoffs/sensitivity
  - Advocating for programs to switch algorithms