CF Newborn Screening in Minnesota

A Clinical Perspective

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Objectives

1. Review the importance of an early diagnosis of CF

2. Review the workflow process of a positive newborn screen for CF from the clinical perspective

3. Identify advantages and challenges of our current practice

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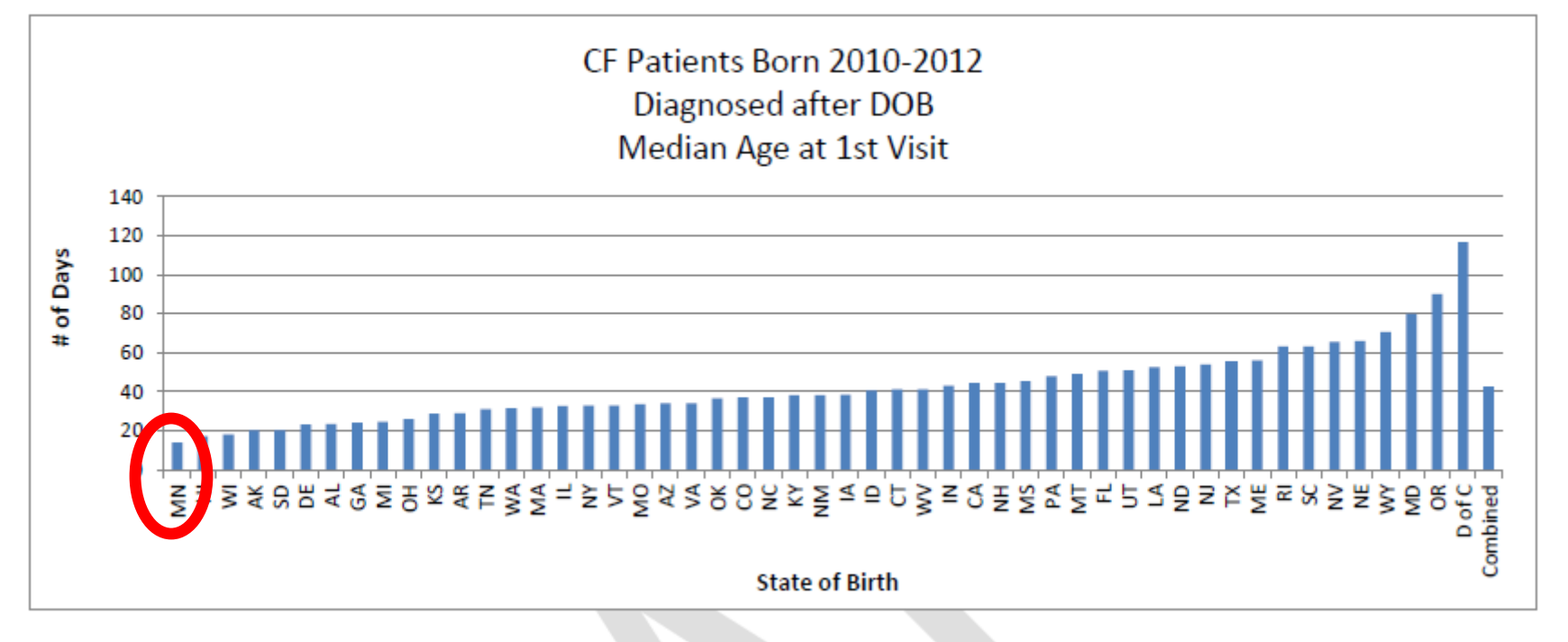
CF Newborn Screening – WHY?

- Malnutrition is typically the earliest manifestation in infants with CF[1]
- Early diagnosis and early treatment with pancreatic enzyme replacement therapy, fat-soluble vitamin supplementation and salt replacement results in improved growth rates in infants with CF[1]
- Well-established correlation between improved nutritional status and pulmonary function as measured by FEV_1 and survival [2-6]

[1]Yen et al. Nutrition in CF, 2015.
[2]Rosenfeld et al. J Am Diet Assoc. 1999
[3]Hankard et al. Horm Res, 2002.
[4]Konstan et al. J Pediatr, 2003.
[5]McPhail et al. J Pediatr, 2008.
[6]Lai HJ et al. Pediatrics, 2009.

What is the dilemma?

Minnesota – Time to first CF Center Visit



<u>Goal</u>: To get infants with a true diagnosis of CF an appointment with an accredited CF Center care team to allow for the initiation of appropriate medical treatment.

Minnesota CF Newborn Screen

- Process is based on an IRT/genetic panel model
- Positive newborn screen is an elevated IRT + at least one CFTR mutation identified by the limited panel
- If two CFTR mutations are identified on the limited panel of 43 mutations, this result is reported
- ***A diagnosis of pancreatic insufficient CF can be identified early*** – CF clinic appointment can be made

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Every process needs a leader…

Genetic Counselors

Amy Powers, CGC

Lynn Schema, CGC

Positive CF NBS – Workflow Process

1. <u>Contact with the CF Center is made by the primary care physician office or the parent</u>
   - Call is received by the Pediatric Clinic Call Center or our CF Administrative Office
   - For all infants < 3 months of age, staff contacts a genetic counselor who responds to the caller within 24 hours

Positive CF NBS – Workflow Process

2. Genetic Counselor contacts physician office or parent to assess patient
   - If two mutations are identified, a CF clinic appointment is made either same or next day
   - If one mutation is identified, growth and overall health is assessed:
     1. If concerns identified, infant followed closely and is often brought in for a sweat test within a week
     2. If no concerns identified, sweat test is scheduled within 3-4 weeks of age

Positive CF NBS – Workflow Process

3. <u>Infant is brought in to Sweat Laboratory for a confirmatory sweat test:</u>
   - Genetic Counselors give our CF Team a heads-up if they are concerned about an infant
   - If sweat test is positive, infant is seen the same day in our CF Clinic
   - If the sweat test is negative, family receives genetic counseling and education

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Advantages

1. Genetic counselors make good quarterbacks!

2. Our Sweat Laboratory can perform sweat test 5 days/week and results are available same day

3. **Logistics make our model work** – our genetic counselors, our sweat laboratory and our CF clinic and multidisciplinary team are all located in the same area – all available 5 days/week

4. Makes the process streamlined for families

Challenges

1. There are a number of moving parts – people must understand their roles and do them

2. Do we need to wait 3-4 weeks to obtain a sweat test or should we do it earlier?

3. We have a QNS protocol in place to minimize our QNS results – must be 2 weeks, at least 8 pounds

4. Is “time to sweat test” the right outcome to measure if the goal is to start therapy ASAP?

Thank you