Provider action sheet for cystic fibrosis (CF)

If the California Newborn Screening (NBS) Program has identified an infant in your care who has a screen-positive result for cystic fibrosis (CF) or a related disorder:

What steps do I need to take?

1. **Consult** with a California Children's Services Special Care Center (SCC) cystic fibrosis specialist. They will ask for your assessment of the infant's current health status and if the infant has any signs of the disorder. The SCC specialist will order confirmatory tests. Further evaluation may be required to make a final diagnosis. Once a definitive diagnosis is made, the goal of treatment is to manage the disease.

2. **Contact** the family to explain the positive newborn screening test result. For information on how best to communicate, consult the **Health Resources & Services Administration (HRSA) Newborn Communication Guide** (https://bit.ly/HRSAGuide or search online for "hrsa" "heritable" "communication")

3. **Review** the accompanying, "Family action sheet for cystic fibrosis" with the family and ensure they understand.

4. **Inform** the family that this result requires more testing and evaluation. Emphasize that you, with NBS Program Area Service Center (ASC) staff and the SCC specialist, will guide them through the next steps of confirmatory testing and follow-up services.

5. **Advise** parents to (1) follow the plan for confirmatory testing, (2) if indicated, begin treatment, and (3) keep their infant's appointments with the SCC specialist. The family should receive services from a multidisciplinary team of specialists, including genetic counseling services.

Clinical information

Cystic fibrosis is caused by mutations in the CF transmembrane conductance regulator (CFTR) gene. The CFTR protein (an ion channel) helps maintain the balance of salt and water throughout the body. CFTR gene mutations, and resultant CFTR protein abnormalities, result in thick, sticky mucus in the lungs, digestive system, and elsewhere in the body.

The disease presentation and severity vary among affected individuals, but common symptoms include salty sweat, chronic cough, frequent infections, and malabsorption leading to poor growth. Treatment for CF may include CFTR modulator drugs, antibiotics, mucolytics, pancreatic enzyme replacement, and airway clearance. Early diagnosis and treatment has led to vastly improved outcomes and life expectancy for people with CF.

Newborn screening can also find infants with CFTR-Related Metabolic Syndrome, or CRMS. These babies should be monitored for symptoms since they may develop cystic fibrosis as they grow old.

Please visit these sites. Search for the site name and "cystic fibrosis" if needed.

- **HRSA** (https://newbornscreening.hrsa.gov/conditions/cystic-fibrosis)
- **GeneReviews** (https://www.ncbi.nlm.nih.gov/books/NBK1250/)

Questions?

For follow-up questions, please call your NBS Program Area Service Center.

For program questions, please email NBS Program staff at NBS@cdph.ca.gov or visit the NBS Program website (www.cdph.ca.gov/NBS).

California Department of Public Health
Newborn Screening Program (www.cdph.ca.gov/NBS)