Cystic fibrosis (CF) is a genetic condition (inherited from an abnormal gene from each parent) that causes mucus to become abnormally thick and difficult to clear; not only in the lungs but also liver, bone, reproductive tract, and more. It is a progressive condition that results in chronic infection and airway damage as well as problems absorbing protein and fat from the intestines.

Until the 20th century, not much was known about the condition except that babies who tasted salty (when kissed) had a life expectancy of only about 6 months.

In 1938, Dr. Dorothy H. Andersen described CF in the literature. Soon after in 1949, several infants with CF were hospitalized for dehydration with low blood sodium (hyponatremia) and it was realized that they were losing salt in their sweat, which lead to more understanding of why the babies tasted “salty.”

In 1989, the specific gene affected was identified, the cystic fibrosis transmembrane regulator (CFTR). Over the years, more than 2,000 genetic mutations have been discovered.

Most CF diagnoses are now made by newborn genetic screening (by serum immunoreactive trypsin) in all 50 states. The sweat test remains the gold standard of diagnosis.

Today, the median predicted life expectancy for a person born with CF is 47 years old; over half of the CF patients in the United States are over 18 years old. The Cystic Fibrosis Foundation aims to have effective treatments for most CF patients within the next 5 years.

For more information or testing for CF, please call UCLA-Mattel Children’s Hospital Cystic Fibrosis program (for adults and children) at (310) 825-5930 ~Dr. Marilyn Woo

More CF information from Dr. Woo at https://www.youtube.com/watch?v=C_PjdZqXYck

New Cystic Fibrosis Treatments

The goal of CF treatment is to improve quality of life and increase life expectancy. Initial treatments were developed to treat the symptoms of CF.

However, in 2012 a new medication, Ivacaftor (Kalydeco) was the first of an new class of medication that modulates the CFTR gene on a cellular level directly on the defective ion channel.

The medication can either help bring the ion channel to the surface of the cell (where it must be to function) or help to keep the ion channel open.

Ivacaftor is an oral once per day medication that was shown to improve lung function, decrease number of lung infections, and improve weight gain. This treatment is patient specific, as it is tailored to work for certain genetic mutations.

Other CFTR modulators have been approved such as Lumacaftor/Ivacaftor (Orkambi) and Tezacaftor/Ivacaftor (Symdeko). These are also specific to age and genetic mutation. More medication is under development. ~Dr. Douglas Li

More CF information from Dr. Li https://www.youtube.com/watch?v=JSviLIYVF18

CF Diagnosis

Two abnormal sweat chloride tests performed at an CF foundation accredited lab, OR

- Two known disease causing and can obtain their medication AND
- Clinical features of cystic fibrosis A genotype test with only one gene copy or unknown CF mutation does NOT mean the patient does not have CF. Cystic Fibrosis is found in ALL races/ethnic groups. ~Dr. Marilyn Woo

More UCLA Pediatric Pulmonology and Sleep Medicine webinars can found online at https://www.uclahealth.org/mattel/pediatric-pulmonology/videos
Meet the Doctors
PROFILE: DR. DOUGLAS LI

Dr. Li is a board certified pediatric pulmonologist. He treats children with all lung disease, and has a special interest in cystic fibrosis and home ventilators.

He serves as the clinical director the Division of Pediatric Pulmonology and the co-director of the UCLA Cystic Fibrosis Program.

His goal is to improve not only the health of individual children, but also the whole pediatric population by serving as the Chair for the California Thoracic Society Pediatrics Committee, a statewide committee that is dedicated to improving the care of pediatric patients through legislative advocacy and patient/family education.

In his free time, Dr. Li enjoys tending to his fruit and vegetable garden, spending time with his dog, and traveling to find good food.

Dr. Li is accepting new patients in Santa Monica and Westwood.

Ask the Docs

DEAR PED PULM: I was just told my baby has cystic fibrosis. I have read about new treatments. How do I know if she can get them? -CF MOM

DEAR CF MOM: The new CFTR modulators work for patients of certain ages with specific gene mutations. As new medications are developed and studied, the set of genes and age requirements are changing. To best determine if a person is eligible, they should check with your CF care team or visit the website below for official information from the CF Foundation. 
https://www.cff.org/Life-With-CF/Treatments-and-Therapies/Medications/CFTR-Modulator-Therapies/

~Dr. Douglas Li

Respiratory Therapy

Common CF nebulized medications

Provide medications in this order:
1. Bronchodilator (such as albuterol) - to prevent bronchospasm, open airways.
2. Hypertonic saline 7% (3-5 mL) - to hydrate thick mucus; remove mucus from airways.
3. Dornase alpha (Pulmozyme®) - to thin the thick secretions.
4. Aerosolized antibiotic for infections

*Note: Steps 1-4 use with appropriate nebulizer if applicable; confirm with your healthcare team.
5. Clean and disinfect reusable nebulizer cup and any device you breathe through after each use per manufacturer instruction. Also bring this equipment to clinic with you to be inspected yearly.

Question or concerns (non-emergent) regarding your respiratory equipment or airways clearance techniques?
Telemedicine visits are available. E-mail: Ubreathe@mednet.ucla.edu or call (424) 259-8904.

~Matthew Dartt, RCP, RRT-ACCS