CYSTIC FIBROSIS FOUNDATION
ADDING TOMORROWS

CLINICAL CARE GUIDE *for* DIAGNOSIS OF CYSTIC FIBROSIS

Clinical care guidelines[1] that clarify diagnostic criteria, including sweat chloride values, have been developed to standardize the diagnosis of infants with positive newborn screening (NBS) results and older patients with CF symptoms.

DIAGNOSTIC DEFINITIONS	
Cystic Fibrosis	CF is diagnosed when an individual has both a clinical presentation of the disease and evidence of CFTR dysfunction.
Cystic Fibrosis Related Metabolic Syndrome (CRMS)/ Cystic Fibrosis Screen Positive, Inconclusive Diagnosis (CFSPID)	Definitions for CRMS and CFSPID have been combined. CRMS/CFSPID applies to **infants** who have a **positive NBS** test for CF AND either: • A sweat chloride value <30 mmol/L and 2 CFTR mutations, at least 1 of which has unclear phenotypic consequences OR • An intermediate sweat chloride value (30-59 mmol/L) and 1 or 0 CF-causing mutations
CFTR - Related Disorder	A monosymptomatic clinical entity associated with CFTR dysfunction that does not fulfill the diagnostic criteria for CF
Avoid terms like "atypical" or "nonclassical" CF: there is no consensus definition of these terms	

1) Farrell PM, White TB, Ren CL, et al. Diagnosis of Cystic Fibrosis: Consensus Guidelines from the Cystic Fibrosis Foundation. J Peds 2017 S4-S15e.1

The diagnosis of CF is primarily based on abnormal CFTR function obtained through sweat chloride testing.

SWEAT CHLORIDE RANGES		
Diagnosis	**≥ 60mmol/L**	A positive newborn screen, clinical features consistent with CF, or a positive family history
Intermediate Range	**30-59mmol/L**	A positive newborn screen, symptoms of CF, or a positive family history, and sweat chloride values in the intermediate range on two separate occasions may have CF. They should be considered for extended CFTR gene analysis and/or CFTR functional analysis.
Unlikely	**≤ 29mmol/L**	A positive newborn screen, and a sweat chloride of less than 30mmol/L indicates that CF is unlikely. Clinical features that may be consistent with CF, a sweat chloride less than 30mmol/L, indicates that CF is less likely. It may however be considered if evolving clinical criteria and/or CFTR genotyping support CF and not an alternative diagnosis.

For newborns greater then 36 weeks gestation and greater than 2kg (>4.4 lbs) body weight with a positive CF newborn screen (NBS) or a positive prenatal genetic test (recommendation 3), sweat testing should be done at 10 days to 4 weeks of age.

For questions, call **1-800-FIGHT-CF** (800-344-4823) or email **info@cff.org**.

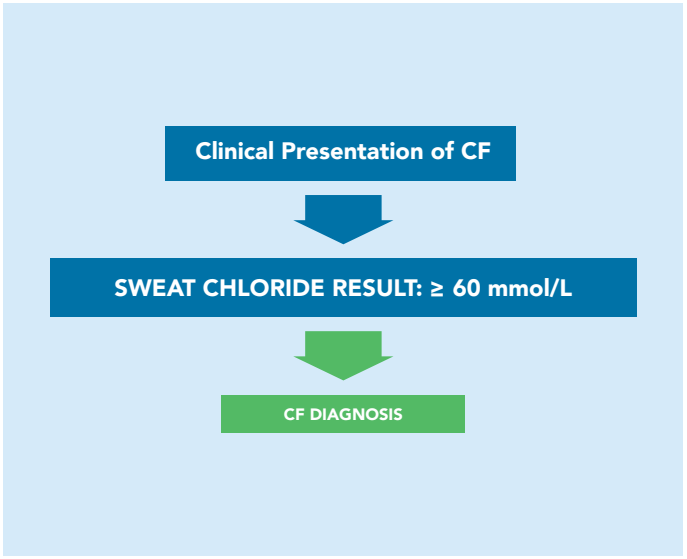
When diagnosing an individual, follow the flow charts to the right with the following definitions in mind:

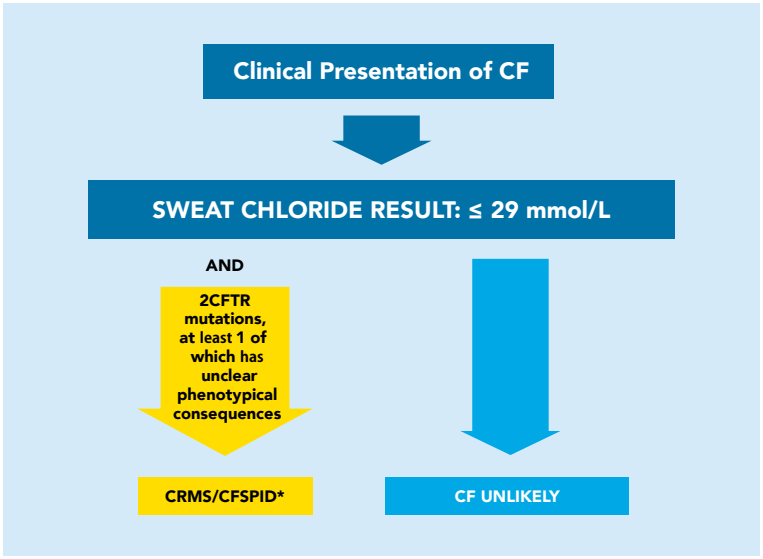
CFTR dysfunction:
Abnormal sweat chloride, presence of CF-causing CFTR mutations, or CF-typical nasal potential difference or intestinal current measurement

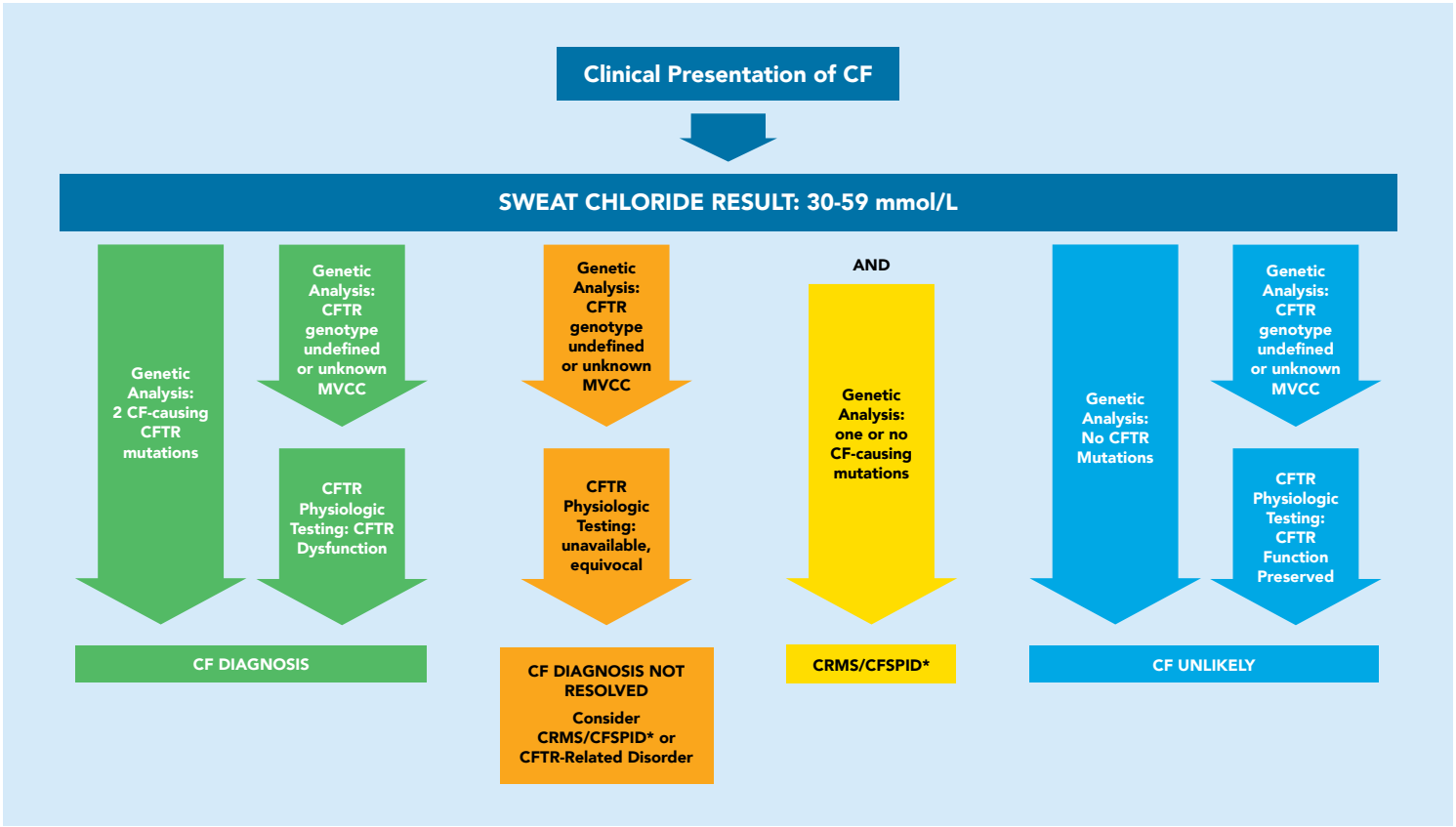
Clinical Presentation of CF includes:
Positive NBS, Signs and/or Symptoms, Family History

MVCC:
Mutations of varying clinical consequence

CFTR2:
A website for the Clinical and Functional Translation of CFTR: cftr2.org







*Positive newborn screen required for CRMS/CFSPID