

Cystic Fibrosis Research News

Citation:

Collaco JM, Blackman SM, Raraigh KS, Corvol H, Rommens JM, Pace RG, Boelle PY, McGready J, Sosnay PR, Strug LJ, Knowles MR, Cutting GR. Sources of Variation in Sweat Chloride Measurements in Cystic Fibrosis. *Am J Respir Crit Care Med*. 2016 Dec 1;194(11):1375-1382.

What was your research question? (50 words maximum)

Chloride sweat tests are used to help determine diagnoses of cystic fibrosis (CF). We estimated potential sources of variation for sweat chloride measurements, including population factors (such as race), testing differences, and different CFTR gene mutations.

Why is this important? (100 words maximum)

Every person has two copies of the cystic fibrosis transmembrane conductance regulator (CFTR) gene. A person must inherit two copies of the CFTR gene that contain mutations, one copy from each parent, to have CF. Chloride, a component of salt, is used to do a sweat test for new diagnoses of CF. People with CF have more chloride in their sweat than people who do not have CF. The chloride concentration in patients' sweat could serve as a useful test of CFTR function in determining how a person responds to potential new CF treatments.

What did you do? (100 words maximum)

More than 2,600 sweat chloride measurements were obtained in more than 1,700 twins/siblings from this study. Measurements of sweat chloride were compared to data such as a patient's CFTR gene mutations, climate, family diet, and genetic information from other genes in the patient's DNA.

What did you find? (100 words maximum)

CFTR gene mutations were found to be the main factor of differences in sweat chloride measurements. Other factors include time (such as issues related to testing on different days), environmental factors (such as climate, family diet),

Cystic Fibrosis Research News

other factors (such as test variability), and individual factors (such as genes other than CFTR in cells and unique exposures). Genes other than CFTR did not play a major role in causing differences in the sweat chloride measurements. For an individual with CF, variation in sweat chloride was mainly caused by variation over time.

What does this mean and reasons for caution? (100 words maximum)

It is essential to have accurate tests of individual medical states, like sweat chloride measurements, to help assess CFTR function in patients with CF undergoing treatment.

What's next? (50 words maximum)

If accuracy of sweat chloride testing can be improved, this measurement can be a valuable test to figure out if therapies directed at CFTR mutations are working well in patients with CF.