

# Observational Study in People with CF with the G551D mutation (GOAL-OB-11)

## Summary

This was a multi-center observational trial to collect clinical information (including height, weight and lung function) as well as specimens (to store and use for future research) in people with CF who have the G551D mutation. The study was initiated at the same time that ivacaftor was approved for use in people with CF and the G551D mutation. The study sought to evaluate response to ivacaftor in the clinical care setting and to evaluate some new potential outcome measures. Additionally biospecimens (blood, urine, and sputum samples) were collected and stored for future research. Researchers for future cystic fibrosis research may use the banked specimens and the health information to develop tests that could be used to help identify additional drugs that can help people with CF. Some participants also had additional procedures performed such as sweat rate testing, ability to clear mucus from the lungs, induced sputum collection, or taking a gastrointestinal pH pill to begin to evaluate the usefulness of these measures for future studies.

## Specimen Information

### Status: Specimens are Available

The primary purpose of this study is to collect specimens from patients with the G-551D mutation who may or may not be prescribed Kalydeco. The specimen collection schedule collects one or two baseline samples (before drug) for those prescribed Kalydeco and 3 after drug samples at varying timepoints. For those not prescribed Kalydeco a baseline sample will be collected and a followup sample will be collected once enrollment for the study has closed. The study also evaluated some novel endpoints including mucociliary clearance, sweat rate, pH Pill and sputum inflammatory markers and microbiome in subsets of enrolled subjects. Sputum samples collected at Visits 1 and 5 have been depleted and are no longer available.

Visit #	Time From Baseline	Specimens Collected
1	-30 Days	Buffy coat, EDTA plasma, Serum, Urine
2	+0 Days	Buffy coat, EDTA plasma, Serum, Urine
3	+1 Months	Buffy coat, EDTA plasma, Serum, Urine
4	+3 Months	Buffy coat, EDTA plasma, Serum, Urine
5	+6 Months	Buffy coat, EDTA plasma, Serum, Urine

### Study Design

<b>Study Type?</b>	Observational
<b>Randomized Study?</b>	No
<b>Placebo Controlled?</b>	No

**Length of Participation** 6 Months

**Number of Study Visits?** 5

### Additional Information

**Phase?** Not Applicable

**Study Sponsor?** Rowe, Steven

**Study Drugs?** N/A

### Eligibility

**Age** 6 Years and Older

**Mutation(s)** One Copy F508del or No Copies F508del

**FEV1% Predicated** No FEV1 Limit

**PA Status** Not Applicable

**Other** Must have the G551D-CFTR mutation on at least 1 allele (any known or unknown mutations allowed in second allele).

## Study Results

### WHAT WE LEARNED:

Study results show that ivacaftor was associated with significant improvements in lung function, body weight and mass, and sweat chloride concentration.

### PRIMARY FINDINGS:

#### EFFECTIVENESS:

This study was conducted between February 2012 and January 2013. Clinical measures and biospecimens were collected from 153 CF patients aged six years and older with at least one G551D mutation and no prior exposure to ivacaftor. Of the 153 participants, 151 were prescribed and received ivacaftor and 133 were followed for 6 months after their first dose. Statistically significant improvements were observed from before taking ivacaftor to after 6 months of treatment in lung function (6.7% improvement in FEV1% predicted [ $p < 0.001$ ]), body weight and body mass, and sweat chloride (mean change from baseline of  $-53.6$  mmol/L to [ $p < 0.001$ ]). There were also significant improvements observed in hospitalization rate ( $P < 0.001$ ) and *Pseudomonas aeruginosa* burden ( $P < 0.01$ ) relative to historical control data. Additionally the experimental outcome measures of mucociliary clearance and gastrointestinal pH were also improved.

#### SAFETY:

Because this was an observational study no safety measures were evaluated.

**CITATION:**

Am J Respir Crit Care Med 2014,DOI 10.1164/rccm.201404-0703OC190(2):175-184

For more information about the results of this study and where it was conducted, visit [ClinicalTrials.gov](http://ClinicalTrials.gov).