



## Cystic Fibrosis Testing

### INTRODUCTION

- Test allows direct DNA analysis of the CFTR gene (CF) to detect 34 specific mutations
- All 23 American College of Obstetrics and Gynecology (ACOG)-recommended mutations detected
- 11 additional mutations included to increase sensitivity
- Reflex R117H for poly T track
- Turn-around time 1 week
- Reported as specific mutations (e.g.,  $\Delta 508$ , R553X, R1162X)



### INDICATIONS

- ACOG recommends screening for women planning pregnancy or during first pregnancy
- Indicated in patients with clinical signs/symptoms of CF, elevated sweat chloride test, or echogenic bowel, for confirmation of diagnosis

### SPECIMEN REQUIREMENTS

4 mL whole blood in EDTA tube

### CLINICAL SIGNIFICANCE

- Approximately 4-5 % of Caucasians carry autosomal recessive mutations for CF
- 30,000 Americans are affected
- Median life expectancy 28 years

### Contact information:

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