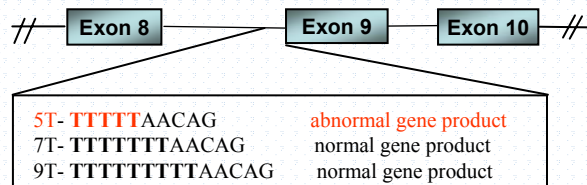


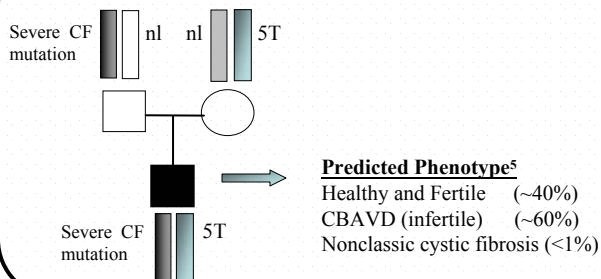
5T quick facts

- Approximately 1 in 10 individuals has the 5T mutation
- The 5T mutation affects splicing of *CFTR* RNA and results in lower than normal amounts of CFTR protein
- The 5T mutation is incompletely penetrant and can be found in healthy individuals, or affected individuals with either CBAVD, or nonclassic cystic fibrosis¹⁻³
- The 5T mutation is associated with the most severe forms of cystic fibrosis when found in *cis* with the R117H mutation⁴
- Testing for 5T in prenatal and population screening is recommended only as a reflex test in cases where the R117H mutation is present

The *CFTR* gene and 5T



Inheritance and consequences of 5T



Testing TG tract length allows for more accurate phenotype prediction^{6,7}

		% with 5T allele who had nonclassic CF or CBAVD*
Standard 5T analysis ⁵	5T	60%
	TG11-5T	10%
New Test for Both TG-T repeats*	TG12-5T	78%
	TG13-5T	> 99%

*Values for TG-T testing were generated from references 6,7. Since most of the affected individuals were men with CBAVD, the above values apply only to men with a CF mutation / 5T genotype



Laboratory Information



DNA Diagnostic Laboratory
 CMSC 10-106
 Johns Hopkins Hospital
 600 N. Wolfe St.
 Baltimore, MD 21287

Phone: 410-955-0483

Fax: 410-955-0484

email: bkarczes@jhmi.edu

Sample Requirement: Minimum of 6cc whole blood in EDTA

Ship to the above address at room temperature with a copy of our requisition form. If more than 36 hours will elapse between time of draw and arrival in the lab, the sample should be refrigerated and shipped with a cold pack. Forms, lab policies, test information and CPT codes are available from our website:

www.hopkinsmedicine.org/dnadiagnostic/

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