

CF PRENATAL SCREENING AND CF NBS PEARLS OF WISDOM

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1

PRENATAL SCREENING

- “Prenatal screening tests can identify whether your baby is more or less likely to have certain birth defects, many of which are genetic disorders. These tests include blood tests, a specific type of ultrasound and prenatal cell-free DNA screening. Prenatal screening tests are usually offered during the first or second trimester. Screening tests can't make a definitive diagnosis. If results indicate an increased risk for a genetic disorder, your health care provider will discuss your options for a diagnostic test to confirm the diagnosis.”¹

1. Mayo Clinic online: www.mayoclinic.org/healthy-lifestyle/pregnancy-week-by-week/indepth/prenatal-testing/art-20045177

2

ACOG RECOMMENDATIONS

- Cystic Fibrosis carrier screening should be offered to all women who are considering pregnancy or are currently pregnant
- Complete analysis of the CFTR gene by DNA sequencing is not appropriate for routine carrier screening
- For couples in which both partners are unaffected but one or both has a family history of cystic fibrosis, genetic counseling and medical record review should be performed to determine if CFTR mutation analysis in the affected family member is available
- If a woman's reproductive partner has cystic fibrosis or apparently isolated congenital bilateral absence of the vas deferens, the couple should be provided follow-up genetic counseling by an OB/GYN or other health care provider with expertise in genetics for mutation analysis and consultation

ACOG committee opinion Carrier screening for Genetic Conditions Number 691 (October 2005)

3

POINTS TO KEEP IN MIND

- Prenatal screening is not 100% sensitive
- Carrier screening is not always offered or completed
- Not all screening tests are created equal
- The risk to be a CF carrier varies by ethnicity

4

MUTATIONS ANALYZED

c.54-5940_273+10250del21kb	c.1973_1985del13insAGAAA	p.R117H	p.R553*
c.262_263delTT	c.1976delA	p.Y122*	p.A559T
c.273+1G>A	c.2012delT	p.G178R	p.R560T
c.273+3A>C	c.2051_2052delAAinsG	p.L206W	p.P574H
c.274-1G>A	c.2052delA	p.F312del	p.R709*
c.313delA	c.2052dupA	p.G330*	p.K710*

**Invitae Core Carrier
Invitae Cystic Fibrosis Test**

Test code: 04714 • 1 gene

Order test

You can customize this test by clicking genes to remove them.

✓ **Primary panel**
1 gene selected

Mutations Analyzed (Continued):

c.1585-1G>A	c.3773dupT	p.V520F	p.S1251N
c.1680-1G>A	c.3889dupT	p.C524*	p.S1255*
c.1766+1G>A	p.E60*	p.G542*	p.W1282*
c.1766+5G>T	p.R75*	p.S549N	p.N1303K
c.1820_1903del84	p.G85E	p.S549R	
c.1911delG	p.E92*	p.G551D	
c.1923_1931del9insA	p.R117C	p.Q552*	

This test was developed and its performance characteristics determined by Esoterix Genetic Laboratories, LLC. It has not been cleared or approved by the Food and Drug Administration. The FDA has determined that such clearance or approval is not necessary.

Urine

5

CF NBS PEARLS

- All baby's with an abnormal CF NBS need a sweat test
 - Yes, even those who had a parent who tested negative for CF on screening
- All baby's with an abnormal NBS for CF should receive genetic counseling
 - These results can have implications for other family members
- Scheduling of the sweat test following an abnormal NBS should always be handled by the genetic counselor
 - Many factors go into play when scheduling the sweat test and counseling

6

MORE PEARLS

- Abnormal NBS results should be given to families in person
- Results typically return the same day the sweat test is completed
- CF NBS is a screening test not a diagnostic test
 - If a baby has clinical signs or symptoms of CF a sweat test should be pursued regardless of the prior NBS results