CF PRENATAL SCREENING AND CF NBS PEARLS OF WISDOM

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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

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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)

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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

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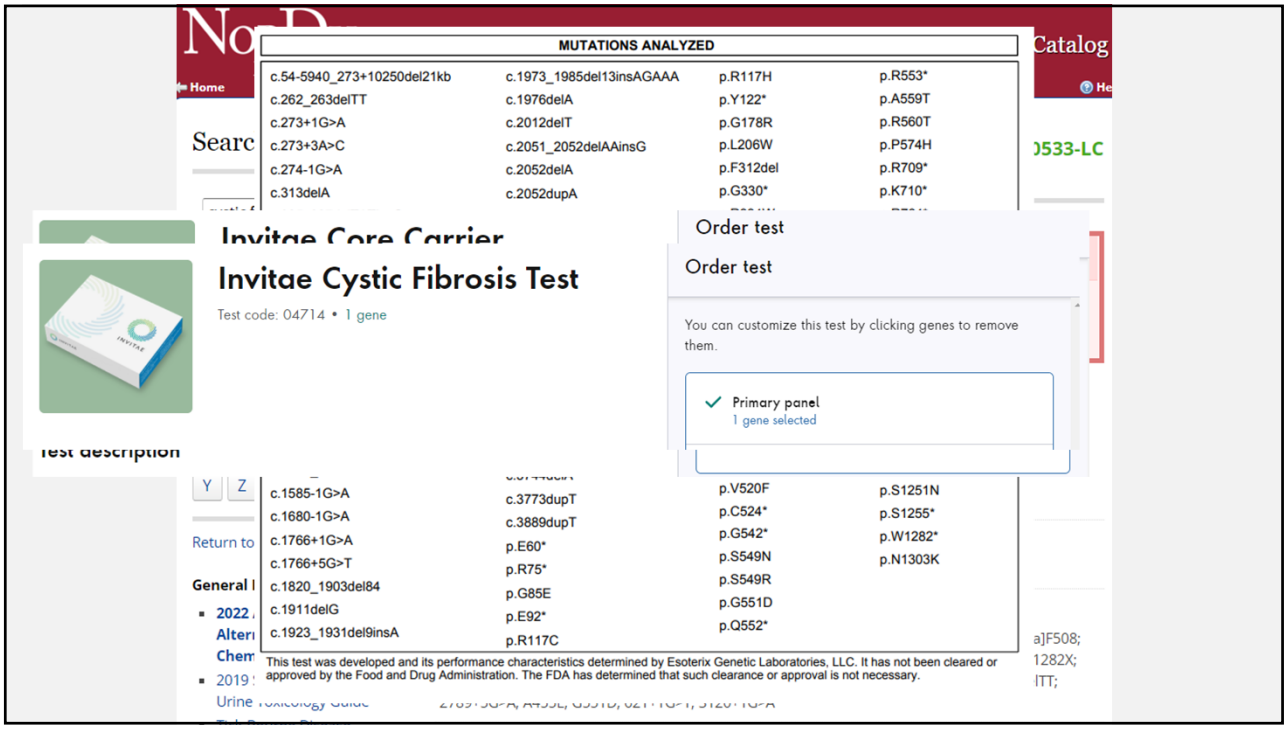

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CF NBS PEARLS

- 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

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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

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