

Integrating Genetic Testing into Practice: Cystic Fibrosis Carrier Screening and the Changing Roles for Clinical and Public Health Professionals

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Abstract

The integration of genetic testing into clinical practice deserves special attention due to the increase in genetic tests available for predicting the risk of human diseases. The recommendations for cystic fibrosis carrier screening provides an example of how the integration of such testing warrants a change in the roles of many disciplines involved including physicians, nurses, genetic counselors, laboratorians and other public health professionals. Consideration must also be given to practice settings and new genetic findings when implementing recommendations that may become standard of care. The focus of this practicum involved using the recommendations from American College of Obstetricians and Gynecologists (ACOG) and the American College of Medical Genetics (ACMG) for cystic fibrosis (CF) carrier screening as a way to explore the genetic testing integration process and the emerging roles of various disciplines, particularly with regard to the laboratory's role.

Practicum Objectives

- **Literature Search:**
 - Identify current genetic testing practices (Physicians, Laboratorians and others)
 - Identify communication process among various disciplines with regard to genetic testing
 - Identify gaps in the current process
 - Review cystic fibrosis carrier screening and ACOG/ACMG guidelines
- **Develop a Model of Emerging Roles**
 - Assess new roles emerging due to cystic fibrosis carrier screening
- **Develop a Survey Instrument**
 - Assess physician knowledge, attitude and use of cystic fibrosis carrier testing

Cystic Fibrosis (CF)

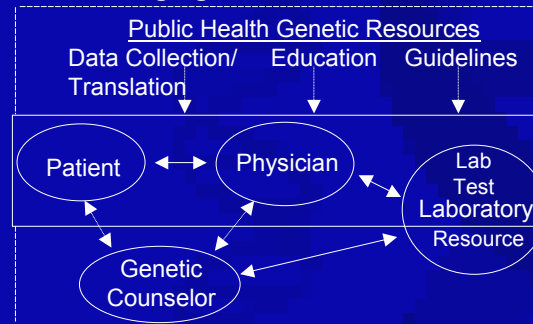
Disease Facts

- Most common autosomal recessive genetic disease
- Incidence of disease varies based on ethnicity
- Carrier risk is 1 in 29 (in the general population)
- Only a few of over 1000 mutations in the CFTR gene have been clinically validated to cause disease

ACOG Recommendations - Carrier Screening

- CF screening *should be offered* to individuals with a family history of CF, reproductive partners of individuals who have CF and couples in whom one or both partners are Caucasian and are planning a pregnancy or seeking prenatal care.
- CF screening *should be made available* to couples in other racial groups who are at lower risk and whom the test may be less sensitive.
- Core pan-ethnic screening panel of 25 mutations - variable detection rate depending on ethnic group

Model Emerging Roles



This model demonstrates the roles that have emerged as a result of CF carrier screening. There is a need for further education to help physicians appropriately assess a patient's risk, order genetic test and interpret and communicate the results. Both the genetic counselors and the laboratorians have an increasing consultative role to help provide this information, introduce additional resources to physicians and help them understand the impacts and limitations of test results.

Emerging Roles due to CF Carrier Screening

Physicians

Practice: Limited knowledge of CF testing, test sensitivity in different ethnic groups, assessing genetic risk and referral to genetic specialists. No significant association between knowledge and discussing the test/test results with patients.

Gaps: Need for clear referral criteria and guidelines to help physicians estimate accurate risks of genetic disease.

Emerging Roles: Physicians need to become familiar with the carrier frequencies and detection rates for different ethnic groups. Also, when ordering a test, providers must be aware of the panel of mutations used by different laboratories.

Laboratorians

Practice: Information necessary for an appropriate laboratory referral (i.e.indication for testing, ethnicity) is not always requested or provided by the laboratories or the referring physician. Laboratory results are often missing critical information for accurate interpretation by physicians.

Gaps: Need for further interpretation of genetic test results by the laboratories performing the test and increased information exchange among physicians and laboratorians.

Emerging Roles: Serve as a consultant and educator to physicians by supplying interpretive results. Need to incorporate requisition forms that require the indication for testing, a family history, and the ethnicity of the patient. Additionally, labs need to indicate which panel of mutations are being tested and their relevance to various populations.

Public Health

Practice: Provide genetic health resources such as information, education, and guidelines. Although there are over 1000 CF mutations, there is limited data of these mutations with respect to clinical disease and variability of expression.

Gaps: Effective method of integrating this information to physicians and other health care professionals.

Emerging Roles: Changing role of how to construct outcome studies to test the clinical validity and clinical utility of genetic tests. Further research needed to understand gene expression and variance for the various CF mutations.

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