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RESEARCH

Public Perceptions of Recessive Carrier Testing in the Preconception and Prenatal Periods

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ABSTRACT

Objective: To explore public perceptions of preconception and prenatal recessive carrier testing.

Design: Qualitative, descriptive.

Setting: Chat rooms located in four websites targeted to those who are pregnant or planning a pregnancy.

Participants: Anonymous comments ($N = 1925$) in online chat rooms.

Methods: The Centers for Disease Control and Prevention's (CDC) Analytic validity, Clinical validity, Clinical utility, Ethical, legal, social implications Model Process (ACCE) for evaluating a genetic test guided this deductive-inductive content analysis.

Results: Participant perceptions of the clinical utility of recessive carrier screening with universal carrier panels are multidimensional. Data analysis revealed four a priori deductive themes present in the data. Secondary inductive analysis produced 20 themes, which exceeded the scope of the CDC's ACCE Model Process for assessing the clinical utility of a genetic test.

Conclusion: Participant perceptions of carrier testing are important to consider in the clinical utility of carrier testing. Participant perceptions of clinical utility vary from those of the CDC's ACCE Model Process and should be considered in evaluation of the clinical utility of recessive carrier testing in the preconception and prenatal populations.

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In reproductive health care, recessive carrier screening is most often used to identify healthy carriers who are at increased risk of having offspring with serious or fatal recessive diseases (Mennuti, 2008). When two individuals who are carriers of the same genetic mutation conceive, there is a 25% chance of them having a child affected by that recessive genetic disorder (Botkin, 2009). Bell et al. (2011) found each individual harbors an average burden of 2.8 known recessive severe pediatric disease mutations. Most carriers of recessive diseases do not typically display symptoms of the disease (Muscular Dystrophy Association, 2014) and could, therefore, be unaware of their genetic risks.

More than 6000 genetic diseases combine to affect 25–30 million people (National Institutes of Health, 2010). While most recessive genetic conditions are individually rare, collectively they affect millions of people globally and account for 10% of pediatric hospitalizations (Bell et al., 2011; Kingsmore et al., 2012). Given these numbers, the World Health Organization (2011) recommended carrier testing for common autosomal recessive diseases, such as hemoglobinopathies.

Genetic carrier screening for autosomal recessive traits, including cystic fibrosis (CF) and hemoglobinopathies, such as sickle cell disease and thalassemia, has become a fundamental component of prenatal and preconception care. In the United States, all pregnant women who receive adequate prenatal care are now offered some form of genetic screening or prenatal diagnosis during the course of their pregnancy (Burke, Tarini, Press, & Evans, 2011). Preconceptual CF screening for people without family history was recommended over a decade ago by the National Institutes of Health, the American College of Obstetricians and Gynecologists, and the American College of Medical Genetics (Grody et al., 2001). Currently, the American College of Obstetricians and Gynecologists (2007, 2011) recommends

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screening all women of reproductive age for CF and those of southeast Asian, African, or Mediterranean descent for hemoglobinopathies.

In the United States, expanded carrier testing options are becoming readily available to individuals for a multitude of single gene conditions. Universal carrier panels (UCPs) are expanded carrier screening tests that screen for multiple genetic mutations that are common across all ethnic groups, therefore making these UCPs applicable to all populations regardless of risk status (Founds, 2014; Latendresse & Deneris, 2015; Srinivasan et al., 2010). Currently, five such panels capable of screening 39 to 167 inheritable conditions are available for clinical use (Counsyl, 2015; GenPath, 2012; LabCorp, 2015; Natera, 2015; Pathway Genomics, 2014).

Background

A review of relevant literature revealed few studies on public perceptions of recessive carrier testing using UCPs in the prenatal and preconception populations with designs at a lower level of evidence. However, current approaches to genetic screening include the practice of using carrier tests to identify couples at risk for having children with inherited genetic conditions (Burke, Tarini, Press, & Evans, 2011; Metcalfe, 2012). Current evidence is generally supportive of carrier screening prior to conception; however, research has been largely limited to single gene conditions. Individuals affected with CF, their parents and relatives, and members of the general public are largely accepting of population screening (Lakeman et al., 2009; Maxwell et al., 2011; Poppelaars et al., 2003). In addition, community members and health professionals are in favor of preconception carrier screening for single gene conditions (Metcalfe, 2012; Watson, Williamson, & Chapple, 1991; Weinreich et al., 2009). Also, Hill, Archibald, Cohen, and Metcalfe (2010) found that women value being offered carrier screening for Fragile X syndrome in the preconception period. Although test uptake, knowledge, and patient attitudes regarding carrier testing with single conditions have been examined (Metcalfe, 2012), there is little evidence regarding perceptions of recessive carrier testing utilizing UCPs in the preconception or prenatal populations. In addition, little is known about how the expansion of carrier screening to UCPs might affect clinical practice and patient uptake (McGowan, Cho, & Sharp, 2013). While UCPs have been available to consumers since 2009, researchers have not examined their clinical utility. Clinical utility refers to the elements that need consideration when evaluating the risks and benefits of introducing a test into routine practice (Haddow & Palomaki, 2010).

As genetic screening options continue to advance, understanding the role of public perceptions is vital to optimizing the clinical utility of UCPs. The goal of preconception and prenatal genetic screening is to provide individuals with information necessary to make informed decisions about their reproduction (Mennuti, 2008). To maximize the clinical utility of UCPs, the provider must understand how individual perspective influences the patient decision-making process. We designed this study to examine public perceptions regarding recessive carrier testing with UCPs and to answer the research question, *What are the themes in public perceptions of the clinical utility of recessive carrier testing?*

Methods

The Centers for Disease Control and Prevention's (CDC, 2010) Analytic validity, Clinical validity, Clinical utility, Ethical, legal, social implications Model Process (ACCE) for evaluation of a genetic test provided a theoretical framework for this study. This evaluation tool includes four components of evaluation of a genetic test (Figure 1): analytic validity, clinical validity, clinical utility, and ethical, legal, and social implications (CDC, 2010). This evaluation tool is composed of 44 standardized questions designed to evaluate each of the components of the genetic test (CDC, 2010). We focused our study on the clinical utility component of the framework, located in the outer ring in Figure 1. Additionally, questions 27–31 of the ACCE tool specifically were used to formulate the deductive nodes for initial data analysis.

The ethics of using publically available chat room data were reviewed carefully prior to submission of this study protocol to an Institutional Review Board for Human Research. The chat room data were publically available existing data not collected for research purposes, submitted voluntarily and anonymously. Other published studies were found using this method (Gilbert & Omisore, 2009; Hoffman-Goetz & Donelle, 2007; Macias, Lewis, & Smith, 2005; Nolan et al., 2008) that

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