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Title:

PHYSICIAN KNOWLEDGE, ATTITUDES, AND PRACTICE REGARDING EXPANDED CARRIER SCREENING

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Objective:

The number of disorders detected by expanded carrier screening (ECS) has increased dramatically over the past few years. Traditionally, carrier screening for single-gene disorders was limited to a few severe diseases in select populations with a high prevalence. Modern advances in genomic platform technology and decreases in cost have made universal screening of a large number of diseases a viable option. Given that technology for genetic testing is rapidly expanding, the knowledge base and attitudes of the physicians who will be employing these tools are increasingly important for patient care. The aim of this study was to better understand individual and practice-related factors that may account for physician utilization of ECS during preconception or prenatal care.

Design:

Prospective

Materials and Methods:

A 20-question online survey was conducted between November 2017 and February 2018. Obstetrician/gynecologists (OB/GYNs) within a single institution were targeted via an email list-serve. The survey assessed current use of ECS, types and number of disorders that can be identified, technologies used, timing for testing, benefits and concerns regarding testing, provision of genetic counseling, and next steps when positive results are obtained. Descriptive statistics were computed for all questions, and chi square tests were performed to compare frequencies according to demographics and practice characteristics.

Results:

A total of 42 responses were collected. Nearly half of respondents (46%) were generalist OB/GYNs (25% practiced GYN only). Respondents practiced in a variety of settings: university hospital (44%); private practice (56%). The majority of respondents (76%) routinely recommended ECS to all patients. Eighty five percent of respondents believed that all patients should be offered ECS, ideally prior to pregnancy. ECS knowledge base was not influenced by provider age or years from training. Compared to providers in an academic setting, providers working in a private setting were more likely to be aware of the correct technology used (82.6% vs. 61.1%, p=0.12) for ECS and the number of diseases it is used to screen (91.3% vs. 66.7%, p=0.04). Compared to generalist OB/GYNs, subspecialists were also more likely to answer these

questions correctly (81.8% vs. 70%, p=0.44; 100% vs. 76.7%, p=0.07). The top 3 potential concerns limiting the use of ECS were financial burden to patients (54%), amount of time/effort spent on counseling patients about results (56%), and amount of time/effort spent on following up on patient results (44%). If both a patient and their partner screened positive, 61% would refer to a genetic counselor, while 24% would refer to a reproductive endocrinologist to discuss in vitro fertilization with preimplantation genetic testing.

Conclusions:

OB/GYNs treating patients preconceptually have the opportunity to be gatekeepers of ECS for their patients. Many OB/GYNs offer ECS to patients and are knowledgeable both benefits and limitations. However, this study demonstrates a need for additional resources to ensure that concerns and knowledge gaps are appropriately addressed and highlights a critical role for genetic counselors as physician educators. ECS has the potential to inform potential parents and to prevent transmission of deleterious mutations to the next generation, provided that obstetricians, gynecologists, genetic counselors, and sub-specialists are aware of and engaged in these scientific advances.