

I. Introduction

Carrier screening began with single-gene screening due to technological and financial limitations. As the number of single-gene screens increased, advancements in screening guidelines and the technologies occurred. These advancements led to the creation of innovative high-throughput platforms, introducing panel-based screening. Expanded carrier screening (ECS) utilizes these panels, screening for hundreds of genes in a single sample. These ECS panels are considered pan-ethnic, decreasing the clinical importance of a patient's ethnic background. However, ECS has a low utilization rate among OB/GYNs (27.1%, Briggs et al., 2018). This low utilization has been attributed to physician lack of comfort and understanding of results, and differing opinions on when to offer ECS (Briggs et al., 2018). In this study, we asked participants to compare their utilization, knowledge, comfort, and attitudes towards ECS in comparison to standard-of-care Cystic Fibrosis (CF) and Spinal Muscular Atrophy (SMA) screening.

II. Methods

Survey

- Created a survey via Qualtrics® to assess areas of interest
- 47 questions total, including closed- and open-ended questions

Participant Recruitment

- Prenatal healthcare providers within the Main Line Health system in Philadelphia, PA
- Emailed providers a link to the Qualtrics® -generated survey

Data Collection

- Responses recorded in Qualtrics® as participants completed survey
- Reported as frequencies and percentages

III. Results

DEMOGRAPHICS

This survey was sent to 153 providers, 43 of whom responded (28.1% response rate). Most of our participants were generalist OB/GYNs (67.4%), and female (81.4%). The age of our participants ranged from 26 to 67 years, and years of practice ranged from less than 5 years to over 30 (Table 1).

UTILIZATION

Utilization of ECS was lower than CF/SMA, with 58.1% and 97.7% of providers reporting use of ECS and CF/SMA screening, respectively. Participants most commonly reported offering CF/SMA screening at a patient's first prenatal visit. For ECS the most common option was not offering the screen at all (Figure 1).

KNOWLEDGE

Most providers reported their knowledge of CF/SMA and ECS as average. However, 93% of the providers ranked their knowledge of CF/SMA average or higher, while only 67.5% of providers ranked their knowledge of ECS within this range (Table 2). Interestingly, when asked to answer a set of questions about ECS, the average score was 88.7%.

COMFORT

For both CF/SMA screening and ECS, providers reported more comfort in reporting negative results compared to positive results. However, it was found that providers are more comfortable discussing positive CF/SMA results than negative ECS results. This was determined from 90.7% of providers reporting being comfortable reporting positive CF/SMA results, and only 65.7% being comfortable reporting negative ECS results (Figure 2).

ATTITUDES

58.1% of the providers selected agreed that it is important for patients to be informed of carrier status to assess risk for a fetus. All others selected "uncertain". 55.8% agreed that positive ECS results led to more complex situations compared to positive CF/SMA. While 53.5% of providers selected agreed that ECS should be offered to all patients, only 46.5% selected "yes" to ECS becoming standard of care (Table 3).

Table 1: Demographics (n=43)

Demographics	Frequency, n(%)
Sex	Male: 8 (18.6%)
	Female: 35 (81.4%)
Profession	Generalist OB/GYN: 29 (67.4%)
	Midwife: 1 (2.3%)
	Nurse Practitioner: 5 (11.6%)
	Physician Assistant: 0 (0%)
	Infertility Specialist: 2 (4.7%)
	MFM Specialist: 6 (14.0%)
Years of practice	<5: 9 (20.9%)
	6-9: 8 (18.6%)
	10-14: 4 (9.3%)
	15-19: 3 (7.0%)
	20-24: 9 (20.9%)
	25-30: 4 (9.3%)
	>30: 6 (14.0%)

Table 2: Provider self-assessment of carrier screening knowledge (n=43)

Self-reported knowledge	CF/SMA, n(%)	ECS, n(%)
High	11 (25.5%)	6 (14.0%)
Above average	11 (25.5%)	4 (9.3%)
Average	218 (41.9%)	19 (44.2%)
Below average	3 (7.0%)	13 (30.2%)
Low	0 (0%)	1 (2.3%)

Table 3: Provider attitudes towards ECS (n=43)

Question	Strongly agree, n(%)	Agree, n(%)	Neutral, n(%)	Disagree, n(%)
It is beneficial for patients to be informed of their carrier risk and to test the partner to determine if the fetus is at risk to be affected with a genetic disorder.	25 (58.1%)	13 (30.2%)	5 (11.6%)	0 (0%)
Positive ECS results lead to more complex situations than positive CF and SMA results.	8 (18.6%)	16 (37.2%)	13 (30.2%)	6 (14.0%)
ECS should be offered to all patients.	11 (25.6%)	12 (27.9%)	12 (27.9%)	6 (14.0%)

Figure 1. Provider reports of when carrier screening is ordered for a patient (n=43)

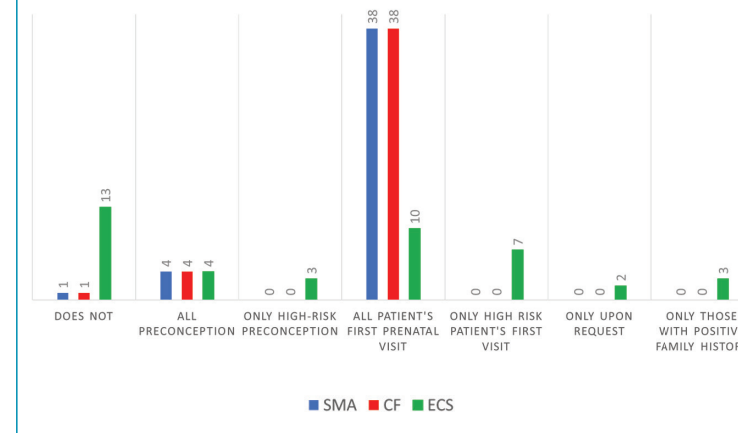
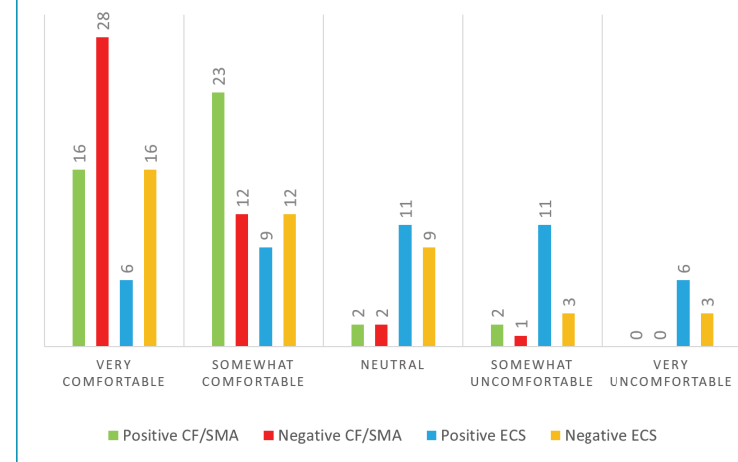


Figure 2. Provider self-assessment on comfort level delivering carrier screening results (n=43)



IV. Conclusions

Ultimately, our study was able to identify the utilization, knowledge level, comfort level and attitudes surrounding ECS for obstetrical providers by comparing uptake with CF/SMA screening.

- The 58.1% of provider utilization of ECS was higher than the 27.1% reported in a previous study (27.1%) (Briggs et al., 2018).
- Providers scored well on our knowledge-based questions after ranking their knowledge as average. Better education for healthcare providers, specifically about onset of conditions screened and decreased importance of ethnicity may increase confidence.
- Low levels of comfort for any result of ECS were seen. This could be explained by issues providers reported: increased time counseling, increasing patient anxiety, post-test counseling concerns, and unfamiliarity with screened conditions
- Over half of the providers were against ECS becoming standard of care. This does not mean that they are against ECS, but rather may indicate that providers are still cautious with how new ECS is and desire more information and understanding before offering to their patients.