Clinical Utility of Expanded Carrier Screening

Katie Johansen Taber, PhD; Kyle Beauchamp, PhD; Gabriel Lazarin, MS, CGC; Dale Muzzey, PhD; Aishwarya Arjunan, MS, MPH, CGC; Jim Goldberg, MD

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Study Objectives

Serious recessive and X-linked conditions affect an estimated 1 in 100 pregnancies.1 However, universal screening for only the two conditions recommended by current guidelines, cystic fibrosis and spinal muscular atrophy (SMA),2-4 misses nearly 70% of carriers of rare diseases and fails to detect up to 94% of pregnancies affected with profound and severe conditions, depending on ethnicity.2-7

Expanded carrier screening (ECS), i.e., testing reproductive partners’ carrier status for a large number of recessive and X-linked conditions without regard to ethnicity, addresses this gap. The American College of Obstetricians and Gynecologists recognizes ECS as an acceptable strategy for carrier screening,6 and with American College of Medical Genetics and Genomics, the Society for Maternal-Fetal Medicine, the National Society of Genetic Counselors, and the Perinatal Quality Foundation, acknowledges the unique benefits and considerations of ECS.8

When ECS is undertaken during the preconception period, results enable interventions to reduce the risk of affected pregnancies, such as in vitro fertilization (IVF) with preimplantation genetic testing for monogenic conditions (PGT-M), use of donor gametes, adoption, avoidance of pregnancy altogether, and if or when pregnancy does occur, prenatal diagnostic testing to determine if the pregnancy is affected. When ECS is undertaken during the prenatal period, results enable prenatal diagnostic testing and informed pregnancy management. Here, we describe the impact of ECS results on planned and actual pregnancy management among couples identified as at-risk by expanded carrier screening (ECS) during the preconception or prenatal period.

Methods

1A. Study design

1B. Survey flow

2. Demographics of study participants

3. Conditions for which ARCs reported their future pregnancies were at risk

4. Actions planned or pursued by ARCs screened preconceptionally

5. Prenatal diagnosis in ARCs screened prenatally

6. Prenatal diagnosis in subsequent pregnancies

7. Top reasons ARCs chose not to undergo prenatal diagnosis

Conclusions

Expanded carrier screening guides pregnancy management and results in fewer births affected with serious genetic conditions.

This study demonstrates the clinical utility of screening for serious conditions which have historically gone undetected before birth.

REFERENCES:

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