Clinical experience of expanded carrier testing for routine preconception and prenatal care

Introduction
Expanded carrier testing is recognized by ACOG as an acceptable testing strategy in women’s healthcare (ACOG Committee Opinion 691, March 2017). We report a large OB/GYN practice’s experience with expanded carrier testing including partner testing uptake and reproductive risk.

Methods
A retrospective analysis of expanded carrier testing ordered at a multi-provider clinic was performed. Panel included >200 disorders, hemoglobin evaluation (hemoglobin electrophoresis and red blood cell indices), and HexA enzyme analysis for Tay-Sachs disease. Indeterminate/gray zone results were excluded. Data analyzed included patient demographics, testing outcomes, and reproductive risks.

Results
- Out of 537 female patients tested, 265 (49.3%) were positive for an autosomal recessive disorder; of those, 201 (75.8%) had partner testing.
- Fourteen couples (7.0%) were at risk to have a child affected with an autosomal recessive disorder.
- Thirty women (5.6%) were carriers of an X-linked disorder.
- Ten patients or partners (1.4%) had two mutations detected, indicating that they may have mild or undiagnosed genetic disorder.

Conclusions
- Implementing an expanded carrier testing protocol including partner testing identifies carriers of many disorders and allows for reproductive risk identification.
- Overall, 8.2% of patients/couples in this cohort were identified to have reproductive risk either due to an X-linked disorder or as carriers of the same autosomal recessive disorder.
- This experience demonstrates the yield of expanded carrier testing when routine testing and partner follow-up is incorporated in a busy obstetrical practice.