Expanded Carrier Screening: Adherence to Panel Criteria and Efficiency of At-Risk Couple Detection

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All authors were employed by Myriad Genetics, Inc. at the time of this study

BACKGROUND

- ACOG lists several criteria that should be met by conditions included on expanded carrier screening (ECS) panels:
  1. carrier frequency of 1 in 100 or greater,
  2. well-defined phenotype,
  3. detrimental effect on quality of life,
  4. cause cognitive or physical impairment,
  5. require surgical or medical intervention,
  6. onset early in life, and
  7. diagnosed prenatally.
- ACOG Committee Opinion 690 names 22 conditions as an "example ECS panel."
- We evaluated 176 genes to determine their consistency with ACOG's criteria.

METHODS

- Criteria related to carrier frequency (1), phenotype (2), and severity (3-6) were evaluated based on published literature.
- Prenatal diagnosis (7) was considered applicable to all conditions.

RESULTS

- Evaluation of 176 genes yielded 37 (referred to throughout as the "guidelines-consistent panel") that were consistent with ACOG's panel design criteria.
- This "guidelines-consistent panel" and the example ECS panel had a similar mean number of criteria met (5.8 and 5.6, respectively) and had a similar proportion of genes that met seven (13.5% and 13.6%, respectively) and six (48.6% and 45.5%, respectively) total criteria (Figure 1).
- Both included all conditions currently recommended for screening. However, the guidelines-consistent panel had higher carrier and at-risk couple (ARC) detection rates (63.0% and 84.6%, respectively) than the example ECS panel (44.7% and 54.7%, respectively), relative to a 176-condition panel (Figure 2).
- The relative ARC rate of the guidelines-consistent panel (31:1) was lower than that of the example ECS panel (34:1), indicating that it more efficiently detects at-risk couples (Figure 3).

Presented at ACOG from April 30-May 2, 2021.

RESULTS

Figure 1. Assessment of ACOG criteria adherence.

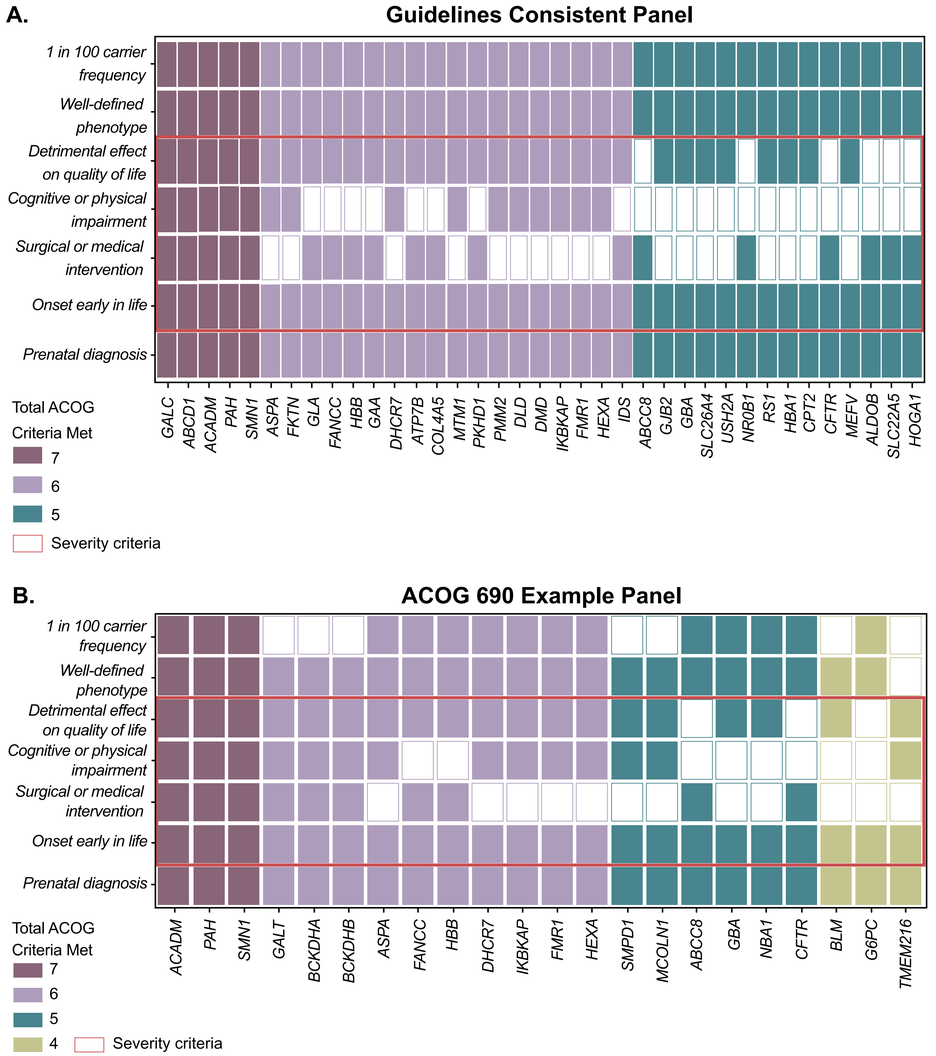


Figure 2. Carrier and ARC detection rates by panel type.

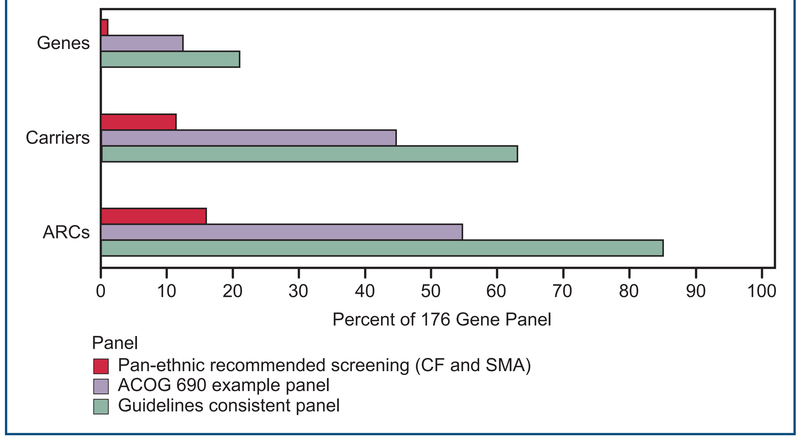
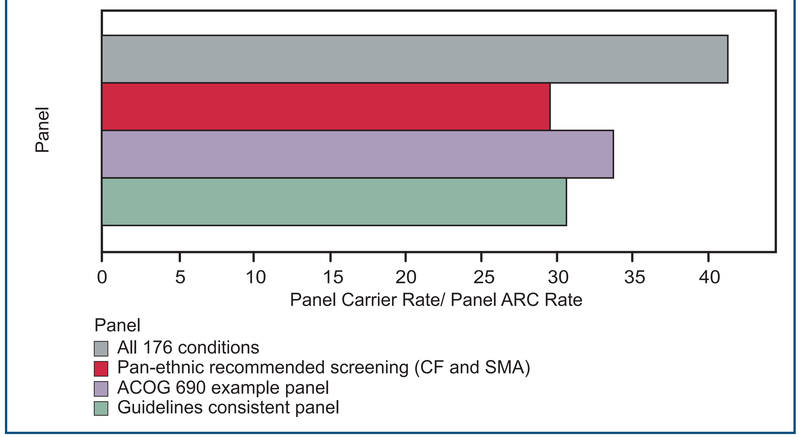


Figure 3. Efficiency of ARC detection by panel type.



CONCLUSION

- A 37-gene ECS panel consistent with ACOG's panel criteria was identified, which more efficiently detects at-risk couples than does the example ECS panel.