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.

● 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).

● 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.

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).

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.