

Some additional points for ESHG's policy include:

- While ESHG recognizes the practicality of offering screening to pregnant individuals, the optimal time to consider carrier screening is prior to conception to provide patients with all reproductive options.
- The primary purpose of carrier screening should be to inform individuals or couples of possible genetic disease risk for future offspring and to allow for reproductive options.
- Expanded carrier screening allows for screening of individuals regardless of their ancestral background, and may increase equity and decrease the odds of stigmatization.
- It should be recognized that the true cost of expanded carrier screening is higher than marketing claims due to follow-up counseling and additional downstream testing.
- The offering of carrier screening should be done thoughtfully to ensure informed decision-making is not compromised.
- The challenges of low genetic literacy, cost and reimbursement, and lack of consistent professional guidelines continue to hinder the offering of carrier screening.
- The clinical validity (the ability of a test to detect or predict carrier status) is uncertain for many conditions due to false-positive variants listed in mutation databases used, incomplete penetrance, and variable expressivity. Therefore priority should be given to screening panels that include a comprehensive set of severe childhood-onset conditions.
- Consent is challenging and should be voluntary and informed. Individuals should know the limitations of testing including:
 - The possibility of false positive/negative results.
 - An individual may be identified as at an increased risk for disease.
- Additional, detailed follow up with a genetics professional, inclusive of psychosocial support, is key to helping interpret and understand results.
- Screening couples at the same time may reduce anxiety and need for additional counseling.
- Pre-test counseling should involve correction of misconceptions about the health status of carriers that may lead to anxiety, as well as limiting false reassurance to individuals who screen negative, given they still have a small residual risk.
- Evidence-based research should continue as expanded carrier screening is initiated to establish acceptable evidence based criteria for its use.