

ACMG lays out criteria for conditions included on any expanded carrier screen and these include:

- Conditions should be ones most individuals at risk would consider prenatal diagnostic testing. Conditions with incomplete penetrance, mild clinical concern, and/or variable expressivity should be clearly noted as such and made optional.
- Adult-onset conditions should be clearly noted, and consent must be obtained. An individual must be able to opt out of obtaining this information.
- Causative genes, mutations, and mutation frequencies should be known for each condition in the population screened. With this, individuals can be provided with meaningful residual risk information. If unknown, this should be clearly noted within patient materials, stressing that results should not be over-interpreted.
- Mutations reported must have a validated clinical association, and further specific literature citations must support that association. This information should be available in both patient and provider materials, such as test results.
- [ACMG's Standards and Guidelines for Clinical Genetics Laboratories](#) should be followed.
- Appropriate patient informational materials should be made available in print and/or through a web site.
- Genetic counseling should be made available to individuals before testing, as desired.
- Genetic counseling is recommended for individuals with positive screen results.