Carrier Screening

Washington State Department of HEALTH

Provider Fact Sheet

Prenatal Testing for Recessive and X-Linked Genetic Conditions

Questions to ask your prenatal and preconception patients

- Is there any history of a genetic condition in your family? If so, and if this is a concern for the patient, it may be best to refer the patient for genetic counseling to ensure that the best carrier test is ordered to identify any specific genetic variants in the family.

- Would you want to learn if you are a carrier for specific genetic conditions?

  Most carriers for recessive and X- linked are not aware they are carriers. There are some tests that can identify people who are carriers for specific genetic conditions.

- Is your reproductive partner available and open to the possibility of carrier screening if you are determined to be a carrier for a genetic condition?

  It is common for people to be a carrier for a genetic condition, however in most cases this does not present any health concerns unless the reproductive partner is also a carrier for the same condition. If both biological parents are a carrier for the same condition, there is typically a 25% or 1 in 4 chances with each pregnancy to have a child with the genetic condition. If you were found to be a carrier, genetic testing would be offered to your reproductive partner.

- Would you consider a diagnostic test, such as amniocentesis or CVS, if you knew that you and your reproductive partner were both carriers for the same genetic condition?

  If you and your reproductive partner are both found to be a carrier for the same genetic condition, the only way to know for certain during pregnancy if the baby is affected or not is through a diagnostic test such as amniocentesis.

  - If you are not pregnant, would you want the option of assisted reproductive technology to screen embryos for genetic conditions?
  - If the answer is no to either of the above two questions, do you feel comfortable waiting until after the baby is born to learn if a genetic condition is present?

- Do you think more information would make you feel prepared?
  - Or does the possibility of more information with the possibility of uncertainty make you anxious?

- Would you prefer not to face the decision of whether or not to have a diagnostic test if you and your reproductive partner are determined to be carriers of a genetic condition?

- Are you confident that carrier screening results wouldn't cause you to do anything differently about your family planning or prenatal testing?

Genetics Task Force Recommendations

- Carrier screening should be offered, not recommended, to all patients. The Genetics Task Force cannot recommend a specific panel or conditions.
- If there is a family history of a genetic condition, consanguinity, or other genetic concerns, referral to a prenatal genetic counselor is recommended.
- The Genetics Task Force recommends carrier screening to those who have not had carrier screening aligned with recent guidelines.
- Most professional societies recommend, at a minimum, cystic fibrosis, spinal muscular atrophy, and hemoglobinopathies screening.
- Larger carrier screening panels (aka expanded carrier screening) are offered by many genetic testing laboratories and is a comprehensive, but not exhaustive, evaluation of reproductive risk for recessive and X-linked disorders. It is up to the providers discretion to decide which panel to use if they choose to.
- Insurance depends on the specific lab/panel chosen as well as the level of insurance coverage.

To learn more about carrier screening and additional resources, visit:

Carrier Screening – Washington State Department of Health
(doh.wa.gov/carrier-screening)

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Created in partnership with the Prenatal Genetics Task Force

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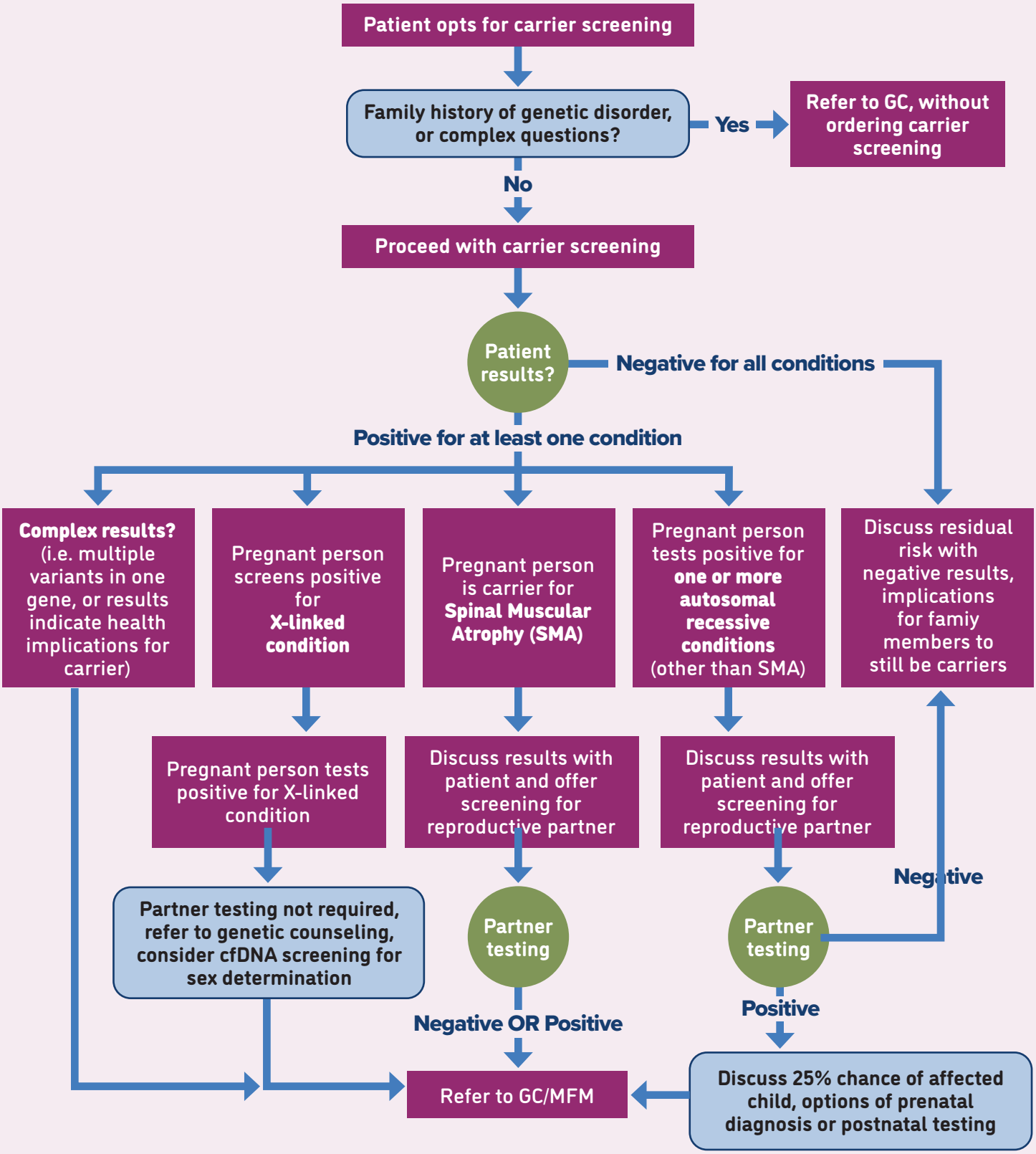
DECISION TREE



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