

## **WHAT PRECONCEPTION GENETIC SCREENING SHOULD BE PERFORMED?**

The challenge of preconception care lies in educating and screening all reproductively capable women on an ongoing basis to identify potential maternal and fetal risks and hazards to pregnancy before and between pregnancies.

Preconception care considerations should include the following factors:

- Undiagnosed, untreated, or poorly controlled medical conditions. These may include diabetes, lupus, hypertension and thyroid disease, asthma, epilepsy, among many others.
- Immunization history with vaccinations offered for women at risk for or susceptible to rubella, varicella and hepatitis B.
- Current medication use with a discussion about fetal safety. This is the best time to change medication to reduce the potential for fetal effects if a better option is available.
- Nutritional issues including optimal weight, food faddisms, and the consumption of a healthy balanced diet. Consideration should be given for folic acid supplementation (1 mg). Higher doses of folic acid (4 mg) are used in women with a history of NTD's or for those on anticonvulsive medication.
- Family history and genetic risk of both parents. The need to screening for sickle hemoglobinopathies, beta or alpha thalassemia, and the "Jewish" panel depends upon ethnicity. Fragile X testing is done when there is a family history of mental retardation or in the patient with a

history of premature ovarian failure desiring fertility treatment. All patients are screened for cystic fibrosis.

- Avoidance of tobacco, substance abuse and other high-risk behaviors.
- Occupational and environmental exposures (i.e. radiation).
- Screening for sexually transmitted diseases with a focus on prevention, including HIV.

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