Screening for Patients with Jewish, Cajun, and French Canadian Heritage

The American College of Obstetrics and Gynecology (ACOG) recommends making screening blood tests available to the above patient populations who are at particular risk for Tay Sachs, Canavan, and Familial Dysautonomia.

There are tests for a number of other medical problems most prevalent in the Jewish population. Additional information about these tests can be found at www.jewishgenetics.org. Although none of these diseases are unique to those of Jewish ancestry, they do occur with higher frequency, making testing most useful for these patients. They are not specifically recommended by the American College of Ob/Gyn, but are recommended by other groups including the American College of Medical Genetics (ACMG).

The ACMG also recommends that all patients be offered testing for Spinal Muscular Atrophy (SMA). However, this is **not** recommended at present by ACOG. Instead, only patients with a family history of SMA or an SMA-like illness are offered testing. SMA is an autosomal recessive neurodegenerative disease that causes atrophy of skeletal muscle and overall weakness. It is caused by a mutation in the survival motor neuron gene (SMA1). The incidence is only about 1 in 10,000 live births, but it is reported to be the leading genetic cause of infant death. Carrier frequencies are 1 in 40 to 1 in 60. There is no effective treatment. The severity of the disease is variable with the most severely affected children dying from respiratory failure within the first two years of life. The most common form is of intermediate severity, with death occurring between the age of 2 and 30. A mild form may cause limited function requiring wheelchair assistance, but may also cause only minor muscle weakness with the ability to walk unaided and a normal life expectancy.