UNC SCHOOL OF MEDICINE

CARRIER SCREENING RECOMMENDATIONS

UPDATED OCTOBER 25, 2019

WHAT IS CURRENTLY RECOMMENDED?

All women should be offered BASIC RECOMMENDED carrier screening for cystic fibrosis (CF), spinal muscular atrophy (SMA) and hemoglobin electrophoresis. Patients should be made aware that more expanded carrier screening is available if desired. Ideally, carrier screening should be offered prior to conception or at the initial prenatal visit. If patients decline carrier screening it should be documented clearly on their Problem List/OB note.

Rescreening is not generally recommended if previously negative.

WHAT ARE THE CORRECT ORDERS IN EPIC?

The correct orders are:

- Lab542 (Cystic fibrosis DNA Panel) – CPT code 81220
- Lab 11165 (SMA Carrier Screen) – CPT code 81329
- Lab5176 (Hemoglobin/Thalassemia Profile UNC) – CPT code 83021
- ICD-10 Code Z13.71 (Testing for genetic disease carrier status)

SHOULD PATIENTS WHO DECLINE SCREENING BE GIVEN ANY OTHER OPTIONS?

Patients can be informed that newborn screening in NC will automatically test for CF and hemoglobinopathies. Newborn screening in NC does not currently check for SMA. However, there are two FDA approved treatments for SMA (a medication and a gene therapy) that dramatically improve outcome for individuals with SMA. However, treatments work best if started within the first week of life. Newborn screening results may take longer than 1 week and does not replace the value of carrier screening. **FOR SMA, PRENATAL DIAGNOSIS CAN IMPROVE LONG TERM OUTCOME.**

Patients can enroll in the EARLY CHECK research study and both screening for SMA and Fragile X will be added to the newborn screen.

WHICH PATIENTS SHOULD BE OFFERED FURTHER TESTING?

Additional carrier screening should be offered in these settings:

- Patient requests expanded carrier screening
- French Canadian/Cajun ancestry – (at increased risk for Tay Sachs)
- Ashkenazi Jewish ancestry – (at increased risk for at least 19 different recessive conditions)
- Consanguinity
- Family history of genetic disease

If referring patients to genetic counseling in this setting **DO NOT** *order CF/SMA in the office as this will be bundled into any larger testing panels which are more cost-effective*.

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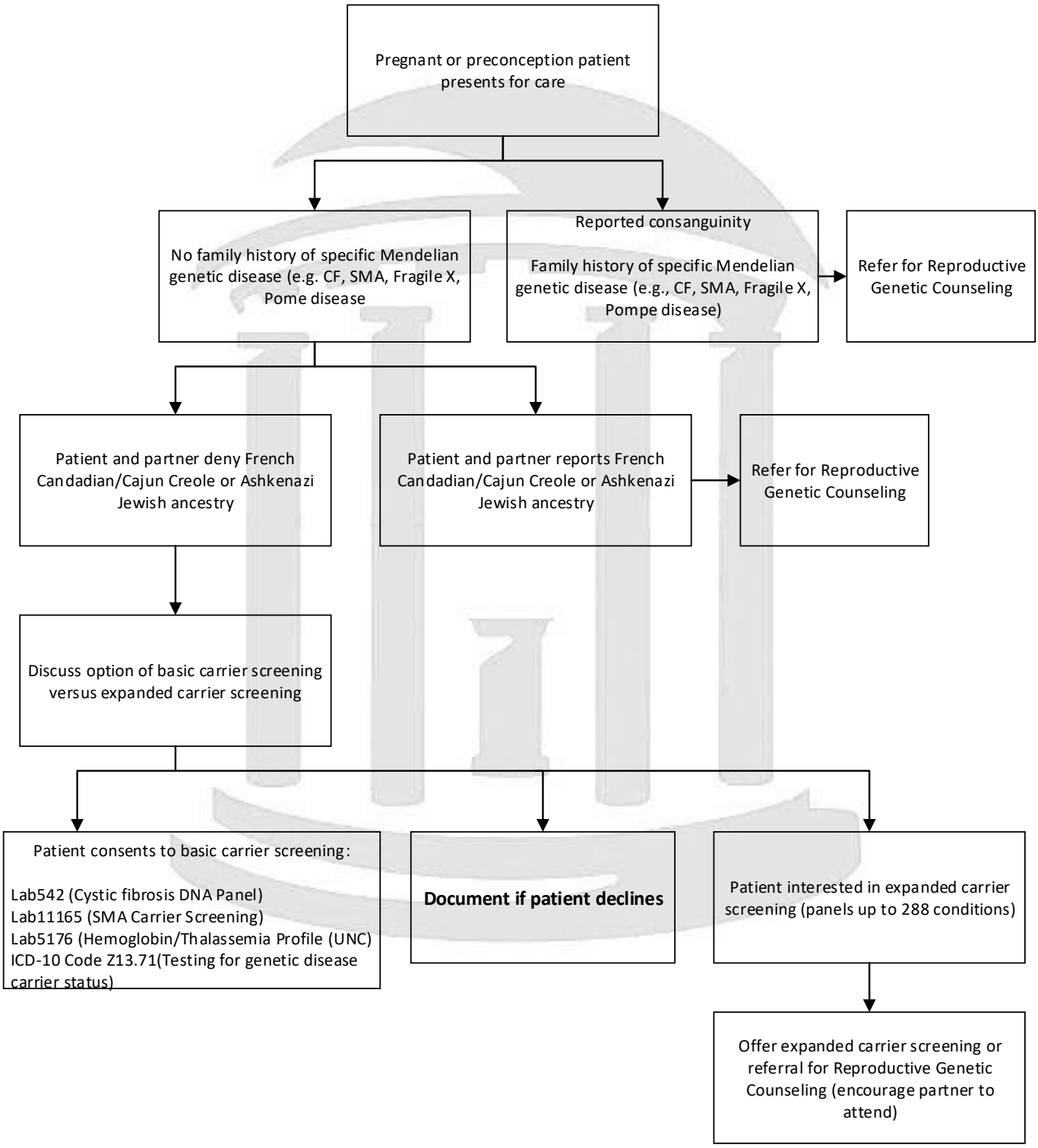

REFERENCES:

1. Carrier screening the age of genomic medicine. Committee Opinion No. 690. American College of Obstetricians and Gynecologists. Obstet Gynecol 2017; 129:E35-40.
2. Carrier screening for genetic conditions. Committee Opinion No. 691. American College of Obstetricians and Gynecologists. Obstet Gynecol 2017;129:e41–55.

Rev. 10/2019 NV/EH

These algorithms are designed to assist the primary care provider in the clinical management of a variety of problems that occur during pregnancy. They should not be interpreted as a standard of care, but instead represent guidelines for management. Variation in practices should take into account such factors as characteristics of the individual patient, health resources, and regional experience with diagnostic and therapeutic modalities.

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