Spinal Muscular Atrophy (SMA) Carrier Screening Overview for Providers (4/2020)

* Spinal muscular atrophy (SMA) is a neurodegenerative disease causing loss of spinal motor neurons resulting in muscle weakness and atrophy.
* SMA is the leading inherited cause of infant death world wide alongside Cystic Fibrosis (CF).
* Autosomal recessive (must receive a defective copy of the gene from both parents).
* SMA carrier frequency 1:40 to 1:50 (somewhat less than CF).
* Caused by deletions in the Survival Motor Neuron (SMN) gene, SMN1.
* Severity modified by a similar gene SMN2.
* Most people inherit one normal SMN1, and one normal SMN2 gene from each parent.
* Both the SMN1 and SMN2 genes are prone to being duplicated, so some people inherit more than one SMN1 and/or SMN2 gene from one orboth parents.
* SMN1 the most important gene. It produces the survival motor neuron protien:
* typical healthy carriers have one copy of a normal SMN1 gene inherited from one parent, and one defective (usually deleted) copy inherited from the other parent.
* UR
* SMN2 produces the same protein as SMN1, but at a much lower level.
* In individuals who have inherited 2 abnormal SMN1 genes, the number of SMN2 genes determine the severity fo te disease.
* The most common scenario in affected individuals is two abnormal/deleted SMN1 genes and 2 normal SMN2 genes. This results in Type 1 SMA in which symptoms begin within 6 months of life, with death 2 years (below).

Spinal Muscular Atrophy Type and Symptoms by SMN2 Number



* 2004-ACOG recommends Universal Cystic Fibrosis carrier screening on pregnant women (or those considering pregnancy) regardless of family history or ethnicity.
* 2017- ACOG recommends Universal Spinal Muscular Atrophy carrier screening on pregnant women (or those considering pregnancy) regardless of family history or ethnicity.
* Currently (4/2020) UR will use the Integrated Genetics SMA carrier screen (materials attached). Call the Reproductive Genetics Counselors at 585-487-3480 with questions. In the future we hope to shift to Mayo Labs so that both CF and SMA can be ordered on the same requisition…stay tuned! The New OB Smartsets all have SMA in the checklist, with a link to the correct Epic order and a Patient Information handout.

This document was sent with several other attachments that should be all you need to start offering SMA screening:

* Integrated Genetics Requisition and Consent; the req is pre-filled out with the test specifics; your staff will have to fill in patient specifics highlighted (outlined to show up on black and white printer).  The Consent is from LabCorp (they bought Integrated Genetics); it is filled out for SMA carrier screen; you'll fill in patient info, and patient and provider sign at bottom where indicated.  Both of these forms should be taken to the UR lab by the patient for blood draw...we make a copy of these to scan into eRecord in case something gets lost (when it does!) before it reaches the Send-out Lab.
* Patient Information Sheet; can be handed out to patients to provide basic info about SMA and carrier screening
* Spinal Muscular Atrophy Provider info; a little more detailed info about the condition and its genetics.
* SMA Normal Report; UR reports will look a little different, but "normal" reports will say "reduced carrier risk"...don't let that through you, this is the way the reports are worded b/c there are some people, depending upon ethnicity, with 2 copies of SMN1 who are carriers.

Again, please do not hesitate to call any of the Reproductive Genetics counselors or geneticists about questions you may have. When the Laboratory is able to switch to the combined Mayo test requisition, the order in the NOB smartest will be updated, and the new requisitions made available.