IWK Health Department of Pathology and Laboratory Medicine
5850/5980 University Ave. PO Box 9700
Halifax, NS
B3K 6R8

Maternal Serum Screening Lab Bulletin

Date: December 23, 2021
To: All physicians & care teams accessing IWK Maternal Serum Screening (MSS) Service and Laboratories referring specimens for MSS analysis
From: Special Chemistry, Department of Pathology and Laboratory Medicine
Re: Immediate suspension of SLOS screening

PLEASE DISTRIBUTE AS APPROPRIATE

We are currently investigating the risk stratification for SLOS (Smith-Lemli-Opitz Syndrome) screening as the rate of positive screens is higher than expected. Initial investigations suggest an issue with the algorithm/calculation used in the risk assessment software and the issue is believed to be confined to specimens tested after December 9, 2021. Risk assessments for Trisomy 21 do not appear to be impacted. Risk assessments for Trisomy 18 remain under investigation and further updates will be provided as necessary.

SLOS is an autosomal recessive condition with an incidence of 1:20,000 to 1:60,000. It is caused by homozygous or compound heterozygous pathogenic variants in the DHCR7 gene. SLOS is characterized by severe developmental delay and multiple congenital anomalies, many of which can typically be detected prenatally on screening ultrasound.

On review of screening performance for SLOS at the IWK over the past 10 years, the positive predictive value is noted to be less than 1%. During that time, all true positive cases have also had abnormal screening ultrasounds.

Given the performance of the screening ultrasound to identify cases of SLOS, and as we continue to investigate to direct a future solution for biochemical screening, results for SLOS risk assessment will be removed from the screening report.

lori.beach@iwk.nshealth.ca
Dr. Lori Beach, PhD, FCACB
Clinical Biochemist
902-470-3814

or

erika.robinson@iwk.nshealth.ca
Ms. Erika Robinson
Manager, Special Chemistry
902-470-8858