

Smith-Lemli-Opitz Syndrome Detection Possible Using Safer Prenatal Testing

Children's Hospital & Res Ctr Oakland



Smith-Lemli-Opitz Syndrome (SLOS) is a congenital multiple anomaly syndrome caused by an abnormality in the production of cholesterol. SLOS affects the development of about 1 out of 20,000–60,000 children and is associated with multiple birth defects and mental retardation.

The challenge in prenatal diagnosis has been the identification of a non-invasive early-pregnancy test that involves detection of definitive and SLOS-specific components. The established SLOS prenatal test involves detecting increased levels of an essential enzyme by testing either a small tissue sample from outside the sac where the baby develops or amniotic fluid samples, each of which involve invasive procedures and pose a risk to the fetus. Typically these tests are performed between 12 to 18 weeks of gestation. Prenatal testing, including molecular genetic testing has only been performed if the parents are “at-risk” for having an SLOS child because of a previous affected pregnancy and/or child, or based on abnormalities detected by ultrasound. A considerable proportion of SLOS cases are not identified until birth.

The new SLOS, developed at Children’s Hospital Oakland Research Institute (CHORI) by Dr. Cedric Shackleton, in

collaboration with Drs. Li-Wei Guo and William K. Wilson of Rice University, provides a reliable and non-invasive procedure for the detection of SLOS by analyzing maternal urine for characteristic components produced by SLOS affected fetuses as early as 11 to 13 weeks' gestation.

“ *Early detection of a SLOS fetus can offer parents the option of making family planning decisions in the most severe cases. In addition, preliminary research on dietary cholesterol supplementation has yielded some promising results for prenatal treatment of SLOS rendering early prenatal diagnosis essential.*

Quest Diagnostics, the nation's leading provider of diagnostic laboratory testing, information and services, has entered into a nonexclusive license agreement with CHORI for the SLOS technology.

For more information visit www.questdiagnostics.com.

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