Mount Sinai Hospital, an internationally renowned 462-bed patient care, teaching and research hospital in Toronto, has implemented a cutting-edge, multidisciplinary prenatal screening program to detect Down Syndrome in the first trimester of pregnancy. Obstetrics, Medical Imaging, Pathology and Laboratory Medicine and Genetics all work collaboratively at Mount Sinai to provide the initial risk estimate and to follow up positive screens with intensive counselling and definitive testing. Mount Sinai is the first site in Canada to offer first-trimester screening (FTS) as an insured service.

Two aspects of the screening program are particularly noteworthy. Firstly, the Center of Excellence in Obstetric Ultrasound (CEOU) at Mount Sinai Hospital, in conjunction with the Fetal Medicine Foundation (FMF) Canada, has become a national leader in developing a quality infrastructure to ensure accurate ultrasound measurement of the nuchal translucency (NT), an important biomarker of Down Syndrome. Secondly, Mount Sinai’s screening laboratory has adopted a novel IT solution, called Waveforms™, to more efficiently obtain and process the complex information required to generate a meaningful screen risk of Down Syndrome.

Screening for Down Syndrome in the first trimester is a new advance that improves upon the performance of the traditional second trimester prenatal screen carried out from 15 to 20 weeks with higher detection rates (DR) and lower false positive rates (FPR). The test combines ultrasound measurement of fluid in the back of the neck of the fetus, visualized as the nuchal translucency or NT, with biochemical analysis of the maternal blood to determine levels of pregnancy-associated plasma protein-A and free beta-chorionic gonadotropin. These along with the maternal age, weight, race and diabetic status are used to calculate the screen risk of Down Syndrome.

The first trimester screening (FTS) program has grown steadily since it was introduced at Mount Sinai Hospital in January of 2002, and has been associated with a number of important clinical benefits including a 30% reduction in the invasive testing rate (amniocentesis) and increased early detection of other chromosome abnormalities and certain birth defects. Patient satisfaction with the test is high due to the availability of earlier results.

A major challenge in implementing first-trimester screening was to ensure the quality of the NT component of the screen. Measuring the NT is not a trivial exercise and sonographers must learn to do it properly through guided training. Research has clearly shown that sonographers obtain the most accurate and consistent results when trained by a judicious mix of didactic education and expert audit of their submitted images according to accepted international standards. Dr. Jo-Ann Johnson of the Division of Maternal Fetal Medicine at Mount Sinai has been instrumental in setting up the training infrastructure across Canada and developing the evaluation tools to assess measurement quality. Thus far, over 1,400 sonographers have enrolled in the certification program.

NT quality has recently become a hot issue, and various professional and government organizations in Ontario including the College of Physicians and Surgeons of Ontario, the Quality Management Program – Laboratory Services affiliated with the OMA and the Ministry of Health and Long Term Care, are working collaboratively to establish and standardize quality criteria and to apply these to the ongoing monitoring of NT images used in the FTS screen.

The second major challenge to the prenatal program at Mount Sinai has been how to obtain the extensive information needed to generate a meaningful risk estimate of Down Syndrome in a timely manner, and then process the information efficiently.
Potential cost efficiencies are substantial and could total hundreds of thousands of dollars annually with widespread adoption by the laboratory. Application of Waveforms™ technology is equally applicable throughout the hospital, especially in Ambulatory service areas where important clinical information/requests are provided by outside physicians. Use of the intelligent “chaperoning” Waveforms™ software will ensure the accurate transfer of information. Mount Sinai Hospital is currently considering expanding the use of Waveforms™ for a variety of requisitions and clinical programs.

With the quality of the NT being addressed and the means in place to efficiently accommodate the extensive information required to calculate the risk of Down Syndrome, the prenatal screening group at Mount Sinai Hospital is looking to extend the first trimester screening program more widely across Ontario and even beyond. Increased availability of first-trimester screening will mean that more women will benefit from more accurate testing carried out earlier in the pregnancy.

First-trimester screening program timeline

For further information on First-Trimester Screening please visit www.fmfcana.com or contact:
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