

The Health of Canada's Children and Youth: A CICH Profile Genetics and Paediatric Health

Section 7 - Emerging Issues

7.1.2 Genetic Screening – Nuchal Translucency Screening in Pregnancy



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Nuchal translucency screening uses ultrasound to screen for Down syndrome, other conditions caused by an extra chromosome (trisomy 13 and 18), and congenital heart defects. It is performed between 11 and 14 weeks of pregnancy. When nuchal translucency screening is done with blood tests, the results are more accurate. Because nuchal translucency is a screening procedure, it provides a risk estimate for developing one of the conditions. A follow-up diagnostic test – such as chorionic villus sampling or amniocentesis – confirms the diagnosis.

The Society of Obstetricians and Gynaecologists of Canada and the Canadian College of Medical Geneticists recommend that nuchal translucency should only be done where the ultrasound staff is specially trained and accredited to do this procedure. Therefore it is not available in all communities across Canada.¹

¹ Joint SOGC-CCMG Clinical Practice Guideline. No. 261 (Replaces No. 187, February 2007). Prenatal Screening for Fetal Aneuploidy in Singleton Pregnancies. J Obstet Gynaecol Can. 2011;33(7):736–50. http://www.sogc.org/guidelines/documents/gui261CPG1107E.pdf