12–14 week scan with emphasis on risk assessment for fetal structural malformations and fetal loss

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Akademisk avhandling
Som med vederbörligt tillstånd av Medicinska Fakulteten
vid Lunds Universitet för avläggande av doktorsgraden i medicinsk vetenskap
kommer att offentligen försvaras i Jubileumsaulan (ingång 59),
Universitetssjukhuset MAS, Malmö
fredagen 23 mars 2007 klockan 9.00.

Fakultetsopponent: Professor Lars-Åke Matsson,
Sahlgrenska Universitetssjukhuset, Göteborg

Organization	Document name
LUND UNIVERSITY Department of Obstetrics and Gynaecology, Malmö University Hospital, Department of Clincal Sciences, Malmö, Lund University.	DOCTORAL DISSERTATION
	Date of issue 23 March, 2007
	Sponsoring organization
Author(s) Maria Westin	

Title and subtitle

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Abstract

The aim was to evaluate first trimester nuchal translucency (NT) measurement for detection and risk assessment of fetal malformations and fetal loss.

39 572 pregnant women were randomised to either a routine ultrasound scan at 12-14 weeks including NT measurement or to a routine scan at 18-20 weeks. Prenatal diagnosis of major congenital heart malformations was compared between the two groups. In the 12-week group 11% of 61 major heart malformations were prenatally diagnosed vs. 15% of 60 in the 18-week group ($p = 0.60$), 7% vs. 15% being detected at the routine scan ($p = 0.15$).

Consecutive fetuses from the 12-week group were studied with regard to the association between increased NT and 1) major heart malformations 2) lethal or serious malformations 3) malformations of at least intermediate severity, 4) perinatal death, 5) miscarriage and 6) termination of pregnancy (TOP). Different NT cut-offs were chosen à priori and tested prospectively. The sensitivity, false-positive rate, positive and negative likelihood ratios of NT ≥3.0 mm with regard to major heart malformation were 9.6%, 0.8%, 12.0 and 0.9. For fetuses with normal karyotype NT ≥ 3.0 mm increased the risk of lethal or severe malformation 15 times, the risk of malformation of at least intermediate severity 8 times, and the risk of TOP 40 times. NT ≥3mm did not significantly increase the risk of miscarriage or perinatal death.

In a low risk population selected from the 12-week group the risk of second trimester miscarriage was estimated. Risk factors for miscarriage were investigated by logistic regression. The miscarriage rate was 0.5%. The number of previous deliveries and miscarriages independently predicted miscarriage: odds ratio, OR, for each previous delivery 1.48, ($p < 0.0001$); OR for each previous miscarriage 1.34, ($p = 0.01$).

Conclusion: For prenatal detection of cardiac malformations, albeit not statistically significant, the 18-week scan policy seems to be superior to the 12-week scan policy. Increased NT elevates the risk of fetal malformation, but NT measurement is not an appropriate screening tool for fetal malformations because of its low sensitivity. In low-risk pregnancies, after an apparently normal fetus has been confirmed at an NT scan, the risk of subsequent miscarriage is approximately 0.5%. This risk is not affected by NT thickness.

Key words: Nuchal translucency, Prenatal diagnosis, Congenital cardiac defect, Malformations, Miscarriage, Risk assessment

Classification system and/or index termes (if any):

Health Sciences, Obstetrics and gynaecology 0380

Supplementary bibliographical information:	Language
Maria.Westin@med.lu.se	English
ISSN and key title: 1652-8220 12-14-week scan	ISBN 978-91-85559-18-3
Recipient's notes	Number of pages 168 / Price
	Security classification