STUDY RELATIONSHIP BETWEEN NUCHAL TRANSLUCENCY THICKNESS AND CONGENITAL HEART DISEASE IN FETUSES

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Introduction

- Congenital heart disease (CHD) - 8/1000 live birth

- CHD is one of the leading causes of death in infants, accounting for nearly 40% of neonatal deaths due to congenital malformations
Introduction

- Measurement fetal nuchal translucency at 11-14\textsuperscript{th} week of gestation is an effective method for screening chromosomal abnormalities.

- If the fetus has increased NT thickness and normal chromosome, this one is still a high risk of stillbirth in the uterus, genetic syndrome or severe congenital defects, where CHD is serious concern.
Introduction

- If NT > 95\textsuperscript{th}, risk of CHD - 1/16 (6\%) - population (NT <95\textsuperscript{th}) - 8/1000.

- Detection rate of CHD based on NT 3-fold increase compared with the traditional indications based on old risk factors: the mother's diabetes, previous child CHD or exposure to toxins during pregnancy …
NUCHAL TRANSLUCENCY

- Nuchal translucency is the sonographic appearance of subcutaneous accumulation of fluid behind the fetal neck in the first trimester of pregnancy.
- The term translucency is used, irrespective of whether it is septated or not and whether it is confined to the neck or envelopes the whole fetus.
- During the second trimester, the translucency usually resolves and, in a few cases, it evolves into either nuchal edema or cystic hygromas with or without generalized hydrops.
- Measure the widest part of the NT
- Place the cursor at the inner boundary of the NT.
- During the scan, more than one measurement must be taken and the maximum one should be recorded.
PATHOPHYSIOLOGY

- Cardiac dysfunction
- Venous congestion in the head and neck
- Altered composition of the extracellular matrix
- Failure of lymphatic drainage
- Fetal anemia
- Fetal hypoproteinemia
- Fetal infection.
<table>
<thead>
<tr>
<th>Nuchal translucency</th>
<th>Chromosomal Defects</th>
<th>Normal karyotype Fetal death</th>
<th>Major fetal abnormalities</th>
<th>Alive and well</th>
</tr>
</thead>
<tbody>
<tr>
<td>&lt;95&lt;sup&gt;th&lt;/sup&gt; centile</td>
<td>0.2%</td>
<td>1.3%</td>
<td>1.6%</td>
<td>97%</td>
</tr>
<tr>
<td>95&lt;sup&gt;th&lt;/sup&gt;–99&lt;sup&gt;th&lt;/sup&gt; centiles</td>
<td>3.7%</td>
<td>1.3%</td>
<td>2.5%</td>
<td>93%</td>
</tr>
<tr>
<td>3.5–4.4 mm</td>
<td>21.1%</td>
<td>2.7%</td>
<td>10.0%</td>
<td>70%</td>
</tr>
<tr>
<td>4.5–5.4 mm</td>
<td>33.3%</td>
<td>3.4%</td>
<td>18.5%</td>
<td>50%</td>
</tr>
<tr>
<td>5.5–6.4 mm</td>
<td>50.5%</td>
<td>10.1%</td>
<td>24.2%</td>
<td>30%</td>
</tr>
<tr>
<td>≥6.5 mm</td>
<td>64.5%</td>
<td>19.0%</td>
<td>46.2%</td>
<td>15%</td>
</tr>
</tbody>
</table>

N=4,767; Snijders et al 1998; Souka et al 1998; 2001; Michailidis & Economides 2001
INCREASED NUCHAL TRANSLUCENCY WITH NORMAL KARYOTYPE

fetal death-spontaneous miscarriage  structural abnormalities  live birth

<table>
<thead>
<tr>
<th>Fetal NT (mm)</th>
<th>3.0-4.4</th>
<th>4.5-5.4</th>
<th>5.5-6.4</th>
<th>&gt;6.5</th>
</tr>
</thead>
<tbody>
<tr>
<td>%</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
INCREASED NUCHAL TRANSLUCENCY AND CHD
Methods

► Objectives:
To define the correlation between NT thickness and CHD.

► Methods:
Case series study of the women with gestational age from 16 to 40 weeks sent from obstetrical hospitals, who has NT thickness $\geq 2.5\text{mm}$.

► Sample size:
Case series study from 01/2008 to 12/2011.
Methods

Criteria for inclusion:
- Patient consent
- > 18 years old
- NT at 11-14\textsuperscript{th} week \(\geq 2.5\text{mm}\)
- Gestational age from 16 to 40 week
- Singleton

Criteria for exclusion:
- No fetal heart activity at admission
- > Twin pregnancy
- Abnormal karyotype
Results & Discussion

- From 01/2008 to 12/2011 there are 219 pregnant women inclus with NT at 11-14\textsuperscript{th} week $\geq$ 2.5mm.
Results & Discussion

**Image Description:**
- A histogram showing the distribution of a variable labeled as "Tuổi Thai" (Thai Age).
- The x-axis represents different age groups, ranging from 16 to 39 years.
- The y-axis represents the frequency of occurrences, ranging from 0 to 50.
- The histogram indicates a peak around 23 years, with a range of 19 to 27 years, labeled as "23 +/- 4 (n=219)."

**Additional Text:**
- "Biểu đồ 2: Phân bố tuổi thai." (Chart 2: Distribution of Thai age.)
Results & Discussion

Fig 3 Distribution of NT

$$NT = 3.9 \pm 1.4 \text{ mm (Min 2.5 Max 9.2 n=219)}$$
## Results & Discussion

<table>
<thead>
<tr>
<th>Condition</th>
<th>N</th>
</tr>
</thead>
<tbody>
<tr>
<td>AVSD</td>
<td>20</td>
</tr>
<tr>
<td>VSD</td>
<td>12</td>
</tr>
<tr>
<td>Fallot</td>
<td>08</td>
</tr>
<tr>
<td>DORV</td>
<td>04</td>
</tr>
<tr>
<td>HLHS</td>
<td>03</td>
</tr>
<tr>
<td>Ebstein</td>
<td>03</td>
</tr>
<tr>
<td>Tricuspid atresia</td>
<td>03</td>
</tr>
<tr>
<td>PS</td>
<td>02</td>
</tr>
<tr>
<td>UV</td>
<td>01</td>
</tr>
<tr>
<td>TR</td>
<td>01</td>
</tr>
<tr>
<td>PAVSD</td>
<td>01</td>
</tr>
<tr>
<td>PAIVS</td>
<td>01</td>
</tr>
<tr>
<td>Truncus</td>
<td>01</td>
</tr>
<tr>
<td>AS</td>
<td>01</td>
</tr>
</tbody>
</table>
Results & Discussion

61 cases with prenatal diagnosis of CHD progression as following:

- Termination of pregnancy: 38 cases
- Postnatal death: 1 case
- Loss to follow up: 4 cases
- Correct diagnosis: 14 cases
Results & Discussion

![Bar chart showing data](chart.png)

*Biểu đồ 4: Phân bố tỷ lệ BTBS theo khoảng mò sau gay (p < 0,001).*
Limitations of our study is the method in the confirmation or exclusion in diagnosis of CHD, we rely on the results of fetal echocardiography of Cardiologist.

Ideally, all of the prenatal results need to be confirmed postnatal examination by the specialist in live births and autopsy in the remaining cases.
Conclusions

- There are strong correlation between increased NT thickness and CHD in fetuses.

- The prevalence of CHD is higher in case of increasing the NT thickness

- All forms of congenital heart disease can be seen in fetus with increased NT

- Indication of fetal echocardiography when NT ≥ 2.5 mm.
Thanks for your attention!