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

Lê Kim Tuyến, MD
Heart Institute in HCMC

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 11-14th 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.

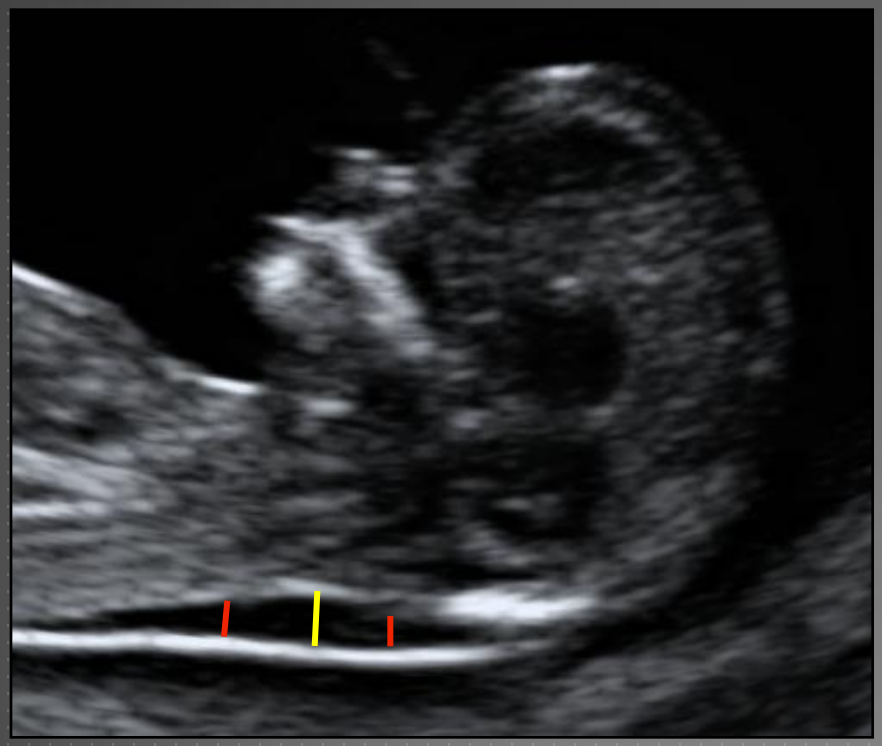
Introduction

- If NT > 95th, risk of CHD - 1/16 (6%) - population (NT <95th) - 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.

NUCHAL TRANSLUCENCY



- 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

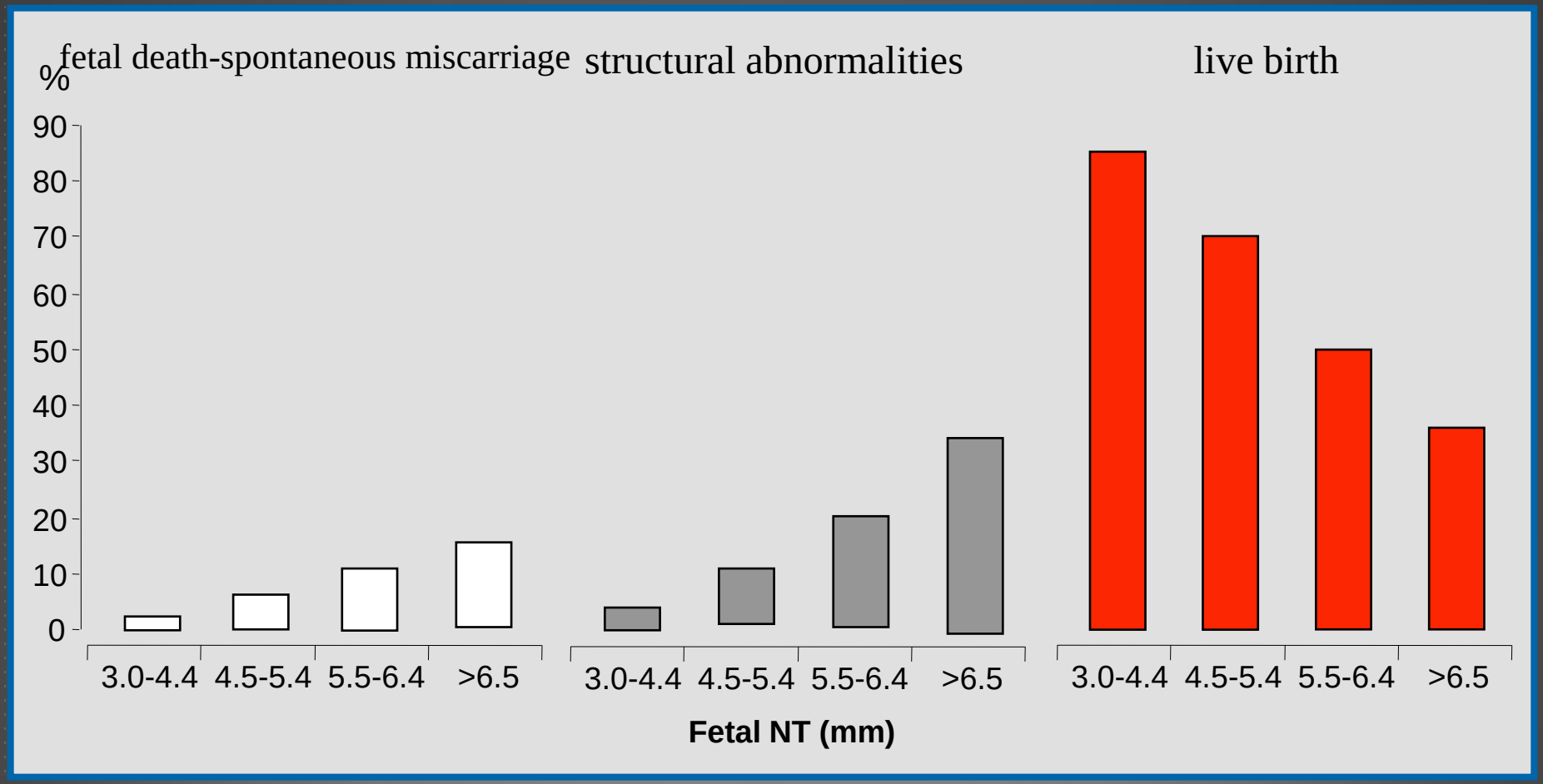
- Cardiac dysfunction
- Venous congestion in the head and neck
- Altered composition of the extracellular matrix
- Failure of lymphatic drainage
- Fetal anemia
- Fetal hypoproteinemia
- Fetal infection.

INCREASED NUCHAL TRANSLUCENCY WITH NORMAL KARYOTYPE

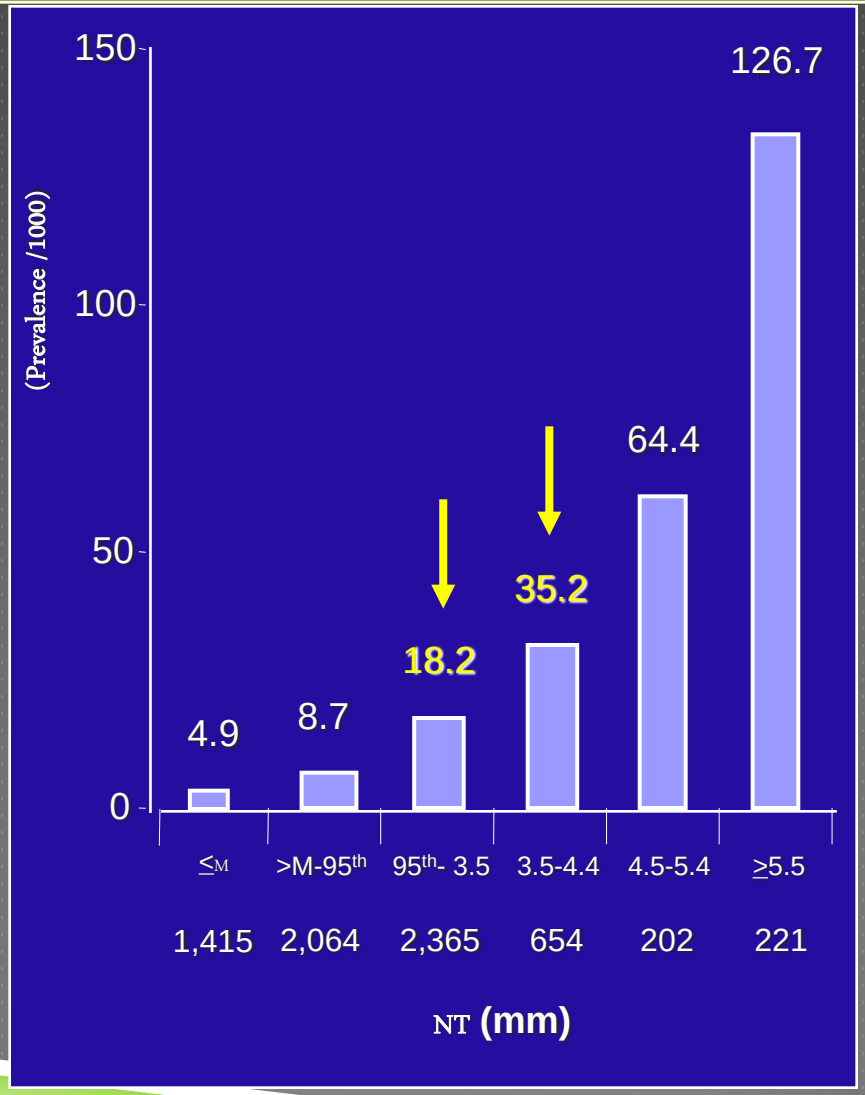
Nuchal translucency	Chromosomal Defects	Normal karyotype		Alive and well
		Fetal death	Major fetal abnormalities	
<95th centile	0.2%	1.3%	1.6%	97%
95th–99th centiles	3.7%	1.3%	2.5%	93%
3.5–4.4 mm	21.1%	2.7%	10.0%	70%
4.5–5.4 mm	33.3%	3.4%	18.5%	50%
5.5–6.4 mm	50.5%	10.1%	24.2%	30%
≥6.5 mm	64.5%	19.0%	46.2%	15%

N=4,767; Snijders et al 1998; Souka et al 1998; 2001; Michailidis & Economides 2001

INCREASED NUCHAL TRANSLUCENCY WITH NORMAL KARYOTYPE



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 $\geq$ 2.5mm.

- Sample size:

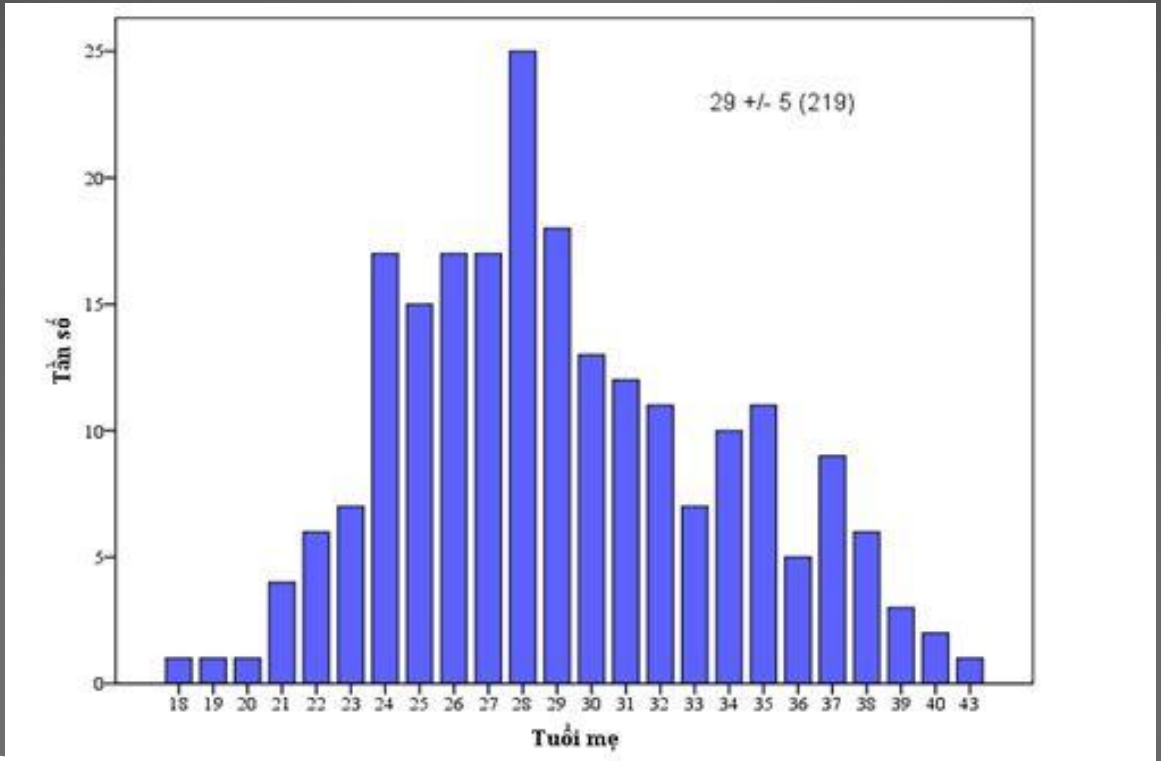
  Case series study from 01/2008 to 12/2011.

Methods

- *Criteria for inclusion:*
  - Patient consent
  - > 18 years old
  - NT at 11-14th week $\geq$ 2.5mm
  - Gestational age from 16 to 40 week
  - *Singleton*
- *Criteria for exclusion:*
  - No fetal heart activity at admission
  - > Twin pregnacy
  - Abnormal karyotype

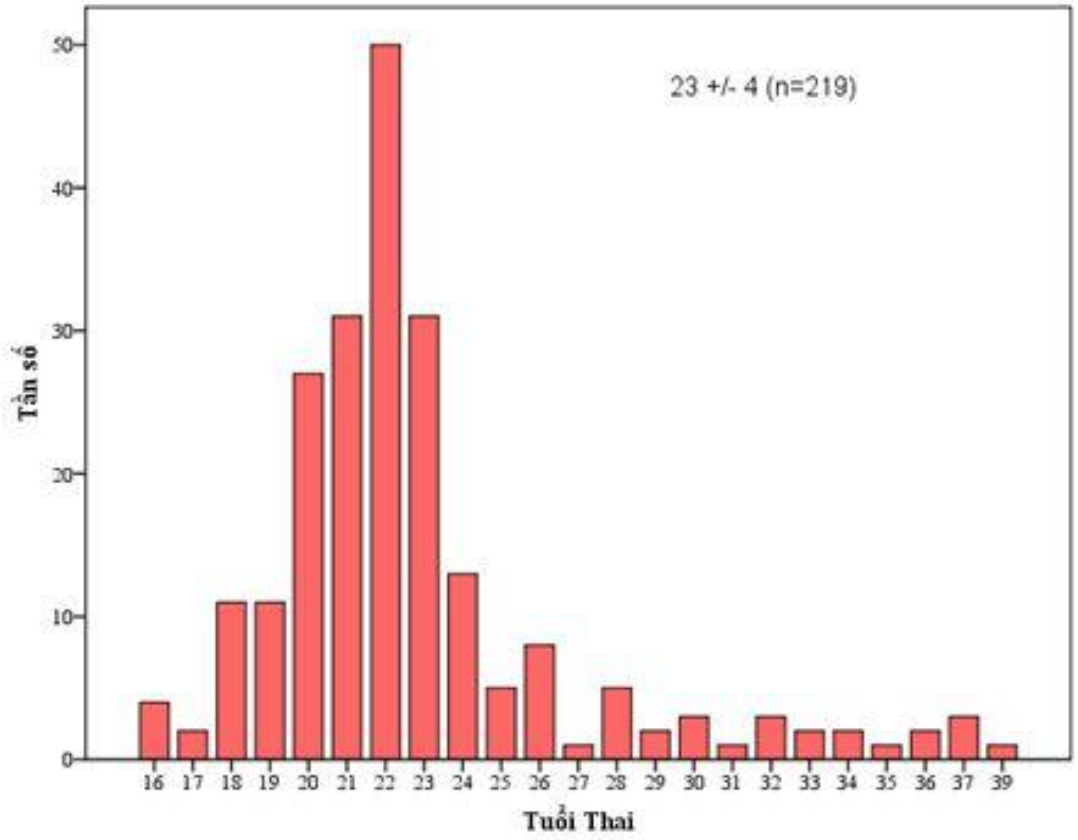
Results & Discussion

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



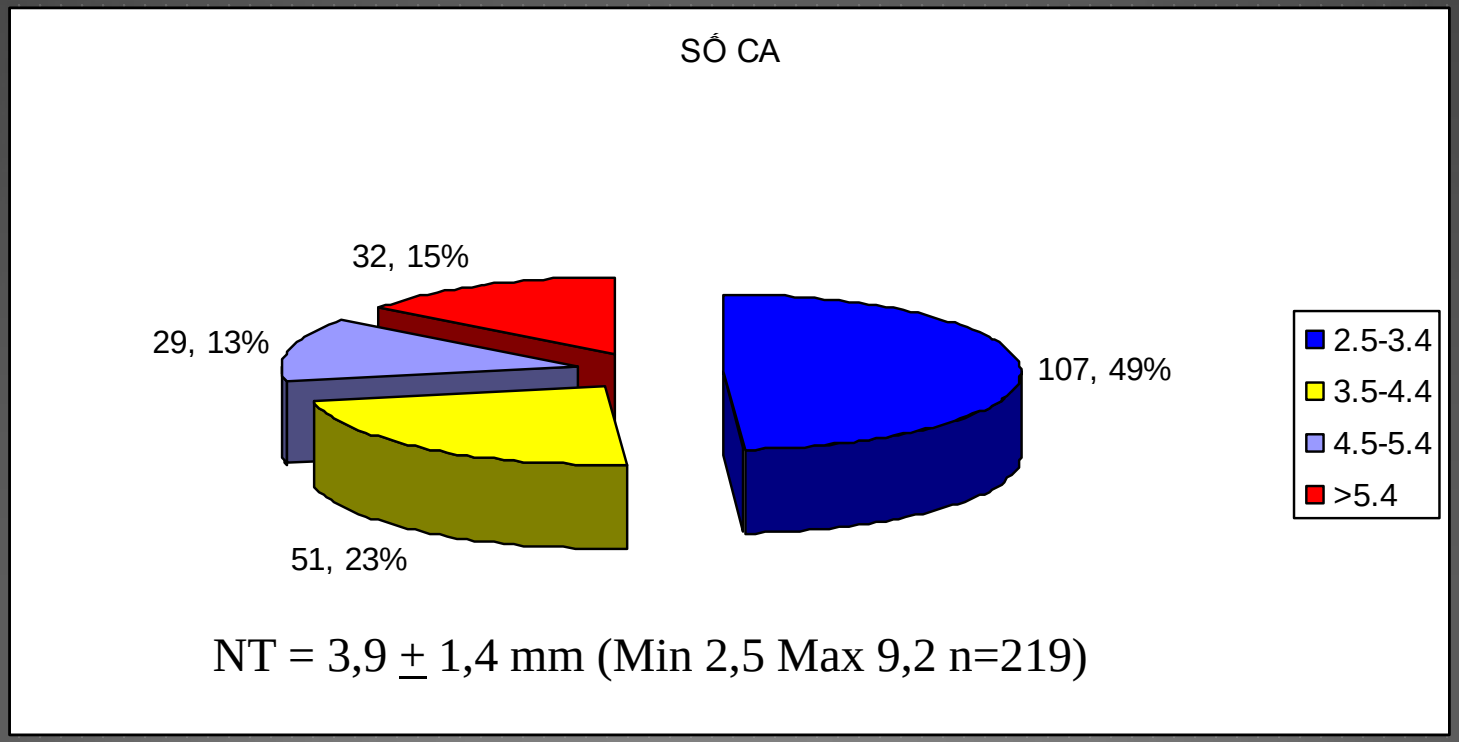
Biểu đồ 1: Phân bố tuổi mẹ.

Results & Discussion



Biểu đồ 2: Phân bố tuổi thai.

Results & Discussion



NT = 3,9 ± 1,4 mm (Min 2,5 Max 9,2 n=219)

Fig 3 Distribution of NT

Results & Discussion

Table 1: List of CHD

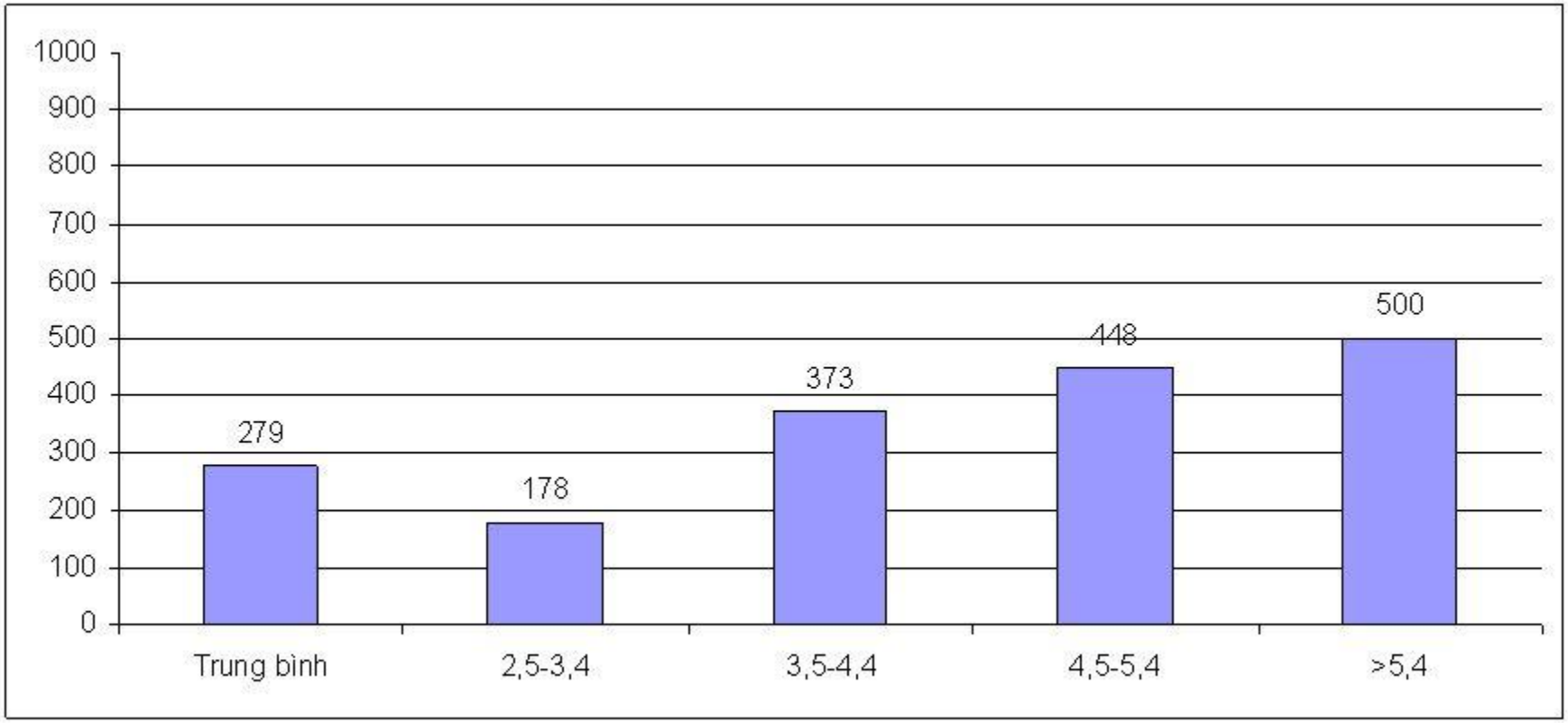
	N
AVSD	20
VSD	12
Fallot	08
DORV	04
HLHS	03
Ebstein	03
Tricuspid atresia	03
PS	02
UV	01
TR	01
PAVSD	01
PAIVS	01
Truncus	01
AS	01

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



Biểu đồ 4: Phân bố tỉ lệ BTBS theo khoảng mờ sau gáy ($p < 0,001$).

- 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 $\geq$ 2.5 mm.

Thanks for your attention!