Increased Nuchal Translucency with Normal Karyotype

New Zealand Maternal Fetal Medicine Network

This guideline was updated in December 2016 by Dr Emma Parry with input from members of the New Zealand Maternal Fetal Medicine Network.

Background

Increased nuchal translucency is associated with chromosomal abnormalities.

It should not be forgotten that increased nuchal translucency with normal karyotype is also associated with a wide range of fetal defects, genetic syndromes and adverse outcomes.

There is unlikely to be a single underlying mechanism for the fluid collection and associated pathology includes cardiac failure, venous congestion, altered extracellular matrix, abnormal lymphatics, infection and anaemia.

Risk of adverse outcome increases with NT thickness.

Objective

To ensure the correct procedure for Nuchal translucency risk assessment is performed

To guide ongoing investigation and management of women presenting with increased nuchal translucency with normal karyotype.

Definition

- Nuchal translucency is the sonographic appearance of subcutaneous accumulation of fluid behind the fetal neck in the first trimester
- This term is used irrespective of septations and whether fluid is confined to the neck or envelopes the whole fetus.
- In the second trimester, this is called nuchal oedema or cystic hygroma.

Normal Values

- Increased nuchal translucency refers to measurement > 95%tile for gestation
- 3.5mm is the 99th%tile and does not change significantly with CRL

Differential Diagnosis

1. Chromosomal (1/3: 75% of which are Trisomy 21 /18)
2. Cardiovascular defects
3. Structural defects: 14.2%
   - diaphragmatic hernia
   - omphalocele
   - thoracic mass
   - skeletal dysplasias
   - body stalk anomaly
4. Congenital infection
5. Metabolic and haematological disorders
6. Syndromes
   - Noonan syndrome
   - Smith Lemli Opitz syndrome
   - Fetal akinesia/hypokinesia
   - Spinal muscular atrophy
   - Congenital adrenal hyperplasia

   And associated Increased risk of miscarriage and perinatal death

Important History

Personal, past obstetric or family history of syndromes, genetic disorders, structural abnormality.

Ultrasound

1. CRL 45-84mm
2. Sagittal section of fetus perpendicular to ultrasound beam
3. Adequate magnification
   - fetal head and upper thorax
   - calliper movement produces 0.1mm change in measurement
4. Fetal neck in neutral position
5. Fetal skin distinguishable from amnion
6. Measurement of subcutaneous translucency between skin and soft tissue overlying cervical spine: without inclusion of skin or soft tissue
7. The horizontal lines of the callipers are placed ON the lines that define nuchal translucency thickness: not IN the line and not IN the nuchal fluid
8. Take more than one measurement and use maximal measurement
9. If there is nuchal cord: measure above and below and use the average

Ultrasound Contd…

Equipment and image quality

1. High resolution ultrasound machine
2. High frequency transducer
3. Manual focussing
4. Appropriate FOV : read/write zoom/depth/narrow sector width
5. Calipers capable of measuring to one decimal point of a mm
6. Zoom magnification capability (magnification should be increased so each increment in the distance between calliper is only 0.1mm)
7. If Sono CT used: switch off harmonic mode and use fundamental mode

Documentation

1. Accurate CRL measurement
2. BPD
3. Both hands, feet, arms and legs
4. Stomach, bladder, abdominal wall, head and spine
5. Nasal bone presence/absence
6. Maternal adnexae

Ongoing Investigation:

3.5 mm is used as the cut off for the requirement for ongoing investigation

1. This is about the 99%tile for nuchal translucency for all CRL
2. It is the balance between detection, increased workload, and available resources

Offer invasive testing (CVS or Amniocentesis) with anuescreen and *array analysis*

<3.5mm with normal karyotype

- Detailed anatomy scan: 2.5% major abnormality
- Confirm nuchal fold thickness not increased
- Careful assessment fetal heart

>3.5mm with normal karyotype

- Early anatomy USS at 16 weeks if CVS performed
- Detailed anatomy scan and Fetal echocardiogram at 20 weeks
- Virology screen if persistent nuchal oedema
- Serial growth scans in 3rd trimester

Prognosis:

Risk of adverse outcome: chromosomal, structural, fetal death

<3.5mm	5%
3.5 – 4.4mm	30%
4.5 – 5.4mm	50%
≥5.5mm	80%

Isolated increased NT with normal karyotype (note this may change with array technology introduction)

< 3.5mm	90% will be normal
3.5-4.4 mm	70% will be normal
4.5–5.4mm	50% will be normal
5.5-6.4mm	30% will be normal
≥ 6.5mm	15% will be normal

Persistent unexplained nuchal oedema at 18-20 weeks

10% chance hydrops, perinatal death, genetic syndrome

3-5% chance neurodevelopmental delay

On-going Management:

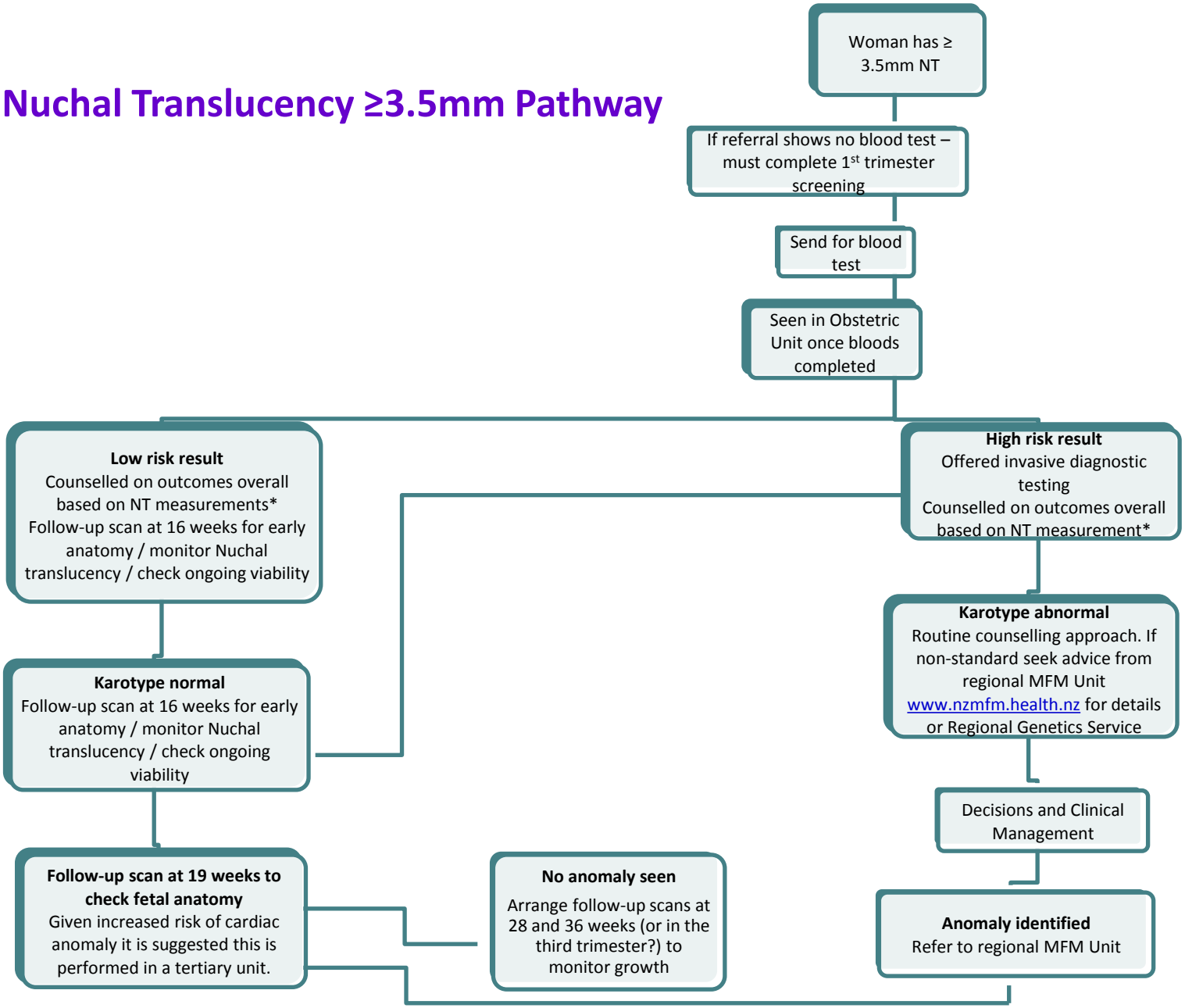
- Increased risk of miscarriage and perinatal death
- Serial USS to assess growth

References:

Nicolaides KH. Nuchal translucency and other first trimester sonographic markers of chromosomal abnormalities. Am J Obstet Gynecol (2004) 191, 45-67

Fetal Medicine Foundation

Nuchal Translucency ≥3.5mm Pathway



*Outcome given Nuchal translucency measurement (adapted from 11-13+^{6} weeks scan by Kypros Nicolaides. *Ref* http://www.fetalmedicine.com/fmf/FMF-English.pdf

Nuchal Translucency (mm)	Chromosomal abnormality (%)	Normal Karotype group			Alive and well given original NT (%)
		Fetal Death (%)	Major Anomaly (%)	Cardiac Anomaly (%)	
3.5-4.4	21.1	2.7	10	3	70
4.5-5.4	33.3	3.4	18.5	7	50
5.5-6.4	50.5	10.1	24.2	20	30
>6.5	64.5	19	46.2	30	15

The outcome data may be better than indicated as in the studies the data was influenced by the number of women with high NT measurements who requested termination of pregnancy.