Mid-trimester Scan and Soft Markers

SKG RADIOLOGY

The mid trimester scan is done to look for fetal abnormalities, and the soft markers are anatomic malformations that are more likely to occur in a fetus with Down's syndrome (trisomy 21) and aneuploidies (including Trisomy 18,13).

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Special interest; Women's imaging.

When should we do the second trimester scan

This should be done at 18-22 weeks.

Why should we do the second trimester scan?

To look for fetal abnormalities, as the prevalence of fetal abnormalities is 2.3%. Of these:

- 63.7% are diagnosed at 18-22 weeks
- 15.7% are diagnosed after 22 weeks
- 20.6% are diagnosed after birth

What are the common anomalies

- CNS anomalies
- Urinary Tract
- Cardiovascular abnormalities

What does the standard anatomical scan include:

Head/skull
- intracranial anatomy

Face - Lips
- Binocular distance and orbits
- Profile, particularly nasal bone

Spine - sagittal views with skin line
- coronal views
- transverse views

Heart - position
- axis
- 4 chamber view

Great vessels
- LVOT
- RVOT
- aortic arch
- ductal arch

Thorax

Diaphragm

Abdomen
- stomach bubble
- anterior abdominal wall
- echogenicity of bowel
- kidneys
- bladder

Umbilical cord
- cord insertion
- presence of 3 vessels

Extremities
- hands
- feet

Why do we do the first trimester scan, maternal serum markers and the second trimester scan?

It is possible to modify an individual's age-related risk of carrying an aneuploid fetus based on first trimester nuchal translucency, maternal serum markers and second trimester scan.

What is a soft marker

A soft marker is defined as an ultrasound finding at 18-22 weeks gestation that increases the risk of the fetus having a chromosomal abnormality (usually Down's Syndrome).

Why do we look for soft markers?

The degree of risk for Down's syndrome, Trisomy 18 and neural tube defect can only be interpreted from the combination of serum screening, and the sonogram.

Soft markers include:

- Nuchal pad > 6mm
- Nasal bone assessment
- Echogenic bowel
- Echogenic foci in fetal heart
- Short humerus <3rd percentile
- Short femur < 3rd percentile
- Pyelectasis (dilated renal pelves) >5mm AP diameter

What is the prior risk?

A patient has a prior risk for Down's syndrome, when she presents for the mid-trimester scan, based on the most accurate prior screening test including nuchal translucency combined with the first trimester maternal serum screen.

What is the new risk?

This can be increased by the presence of isolated soft markers using the formula
New risk=prior risk x likelihood ratio

Simplified likelihood ratio

Marker	Likelihood ratio
Nuchal fold	17x(8-38)
Echogenic bowel	6.1x(3-12.6)
Short humerus	7.5x (4.7-12)
Short femur	2.7x(1.2-6.0)
Hypoplastic nasal bone	83x
Echogenic focus	1

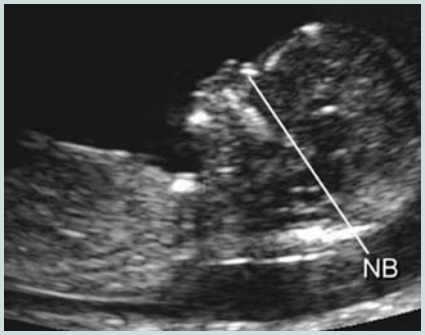

Fig 1.

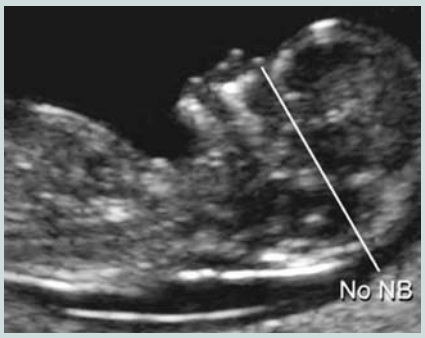

Fig 2.

Comparison: Mid-trimester scan, soft marker assessment. Fig 1: Nasal bone (NB) is present. Fig 2: Nasal bone absent i.e. soft marker.

Management of new risk ratio

If the new risk is greater than 1:250, then the patient should be offered genetic counselling and amniocentesis.
She should also be offered karyotyping if:

1. The nuchal pad alone is increased
2. Finding any 2 soft markers
3. A soft marker in a woman who is otherwise high risk
4. There is a hypoplastic or absent nasal bone

SUMMARY

- All soft markers should be taken in the context of prior risk.
- A new adjusted risk should be calculated.
- If this puts the fetus into high risk (1:250), then genetic counselling and amniocentesis should be offered.