



# Ultra sonographic Soft markers

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# Absent or hypoplastic Nasal Bone

in the **midsagittal fetal profile** with angle of isonation close to 45 or 135, absent or shorter than 2.5mm is significant.

- Sensitivity of **absent NB alone** for detecting **trisomy 21** is **65%**, with a **FPR** of **0.8%**.
- **NIPT** or **Amniocentesis** should be **considered** for **karyotype**





# Echogenic bowel

- Bright lesion in the fetal abdomen with echogenicity similar to **surrounding bone**.
- Aneuploidy 10%
- TORCH infection up to 10%
- Meconium ileus,
- Cystic fibrosis, 15-40%
- Placental hemorrhage, swallowed blood,
- Normal variant





# Echogenic bowel

- Likelihood ratio is **5.5-6.7** for Down syndrome
- Noninvasive prenatal testing (**NIPT**) should be considered for karyotype
- Serial ultrasound examinations to monitor fetal growth, amniotic fluid, with **elevated AFP** is associated with **poor outcomes** [IUGR or IUFD].





# Ventriculomegaly

Ventriculomegaly is mild if the atrial diameter is between 10 - 15 mm and severe if  $>15$  mm

**Detailed anatomic survey** Fetal neurologic scan to look for additional anomalies and **Fetal echocardiogram.**

In isolated ventriculomegaly there is a **4 -fold increase** in risk of trisomy 21.

Genetic counseling **NIPT** or **Amniocentesis.**





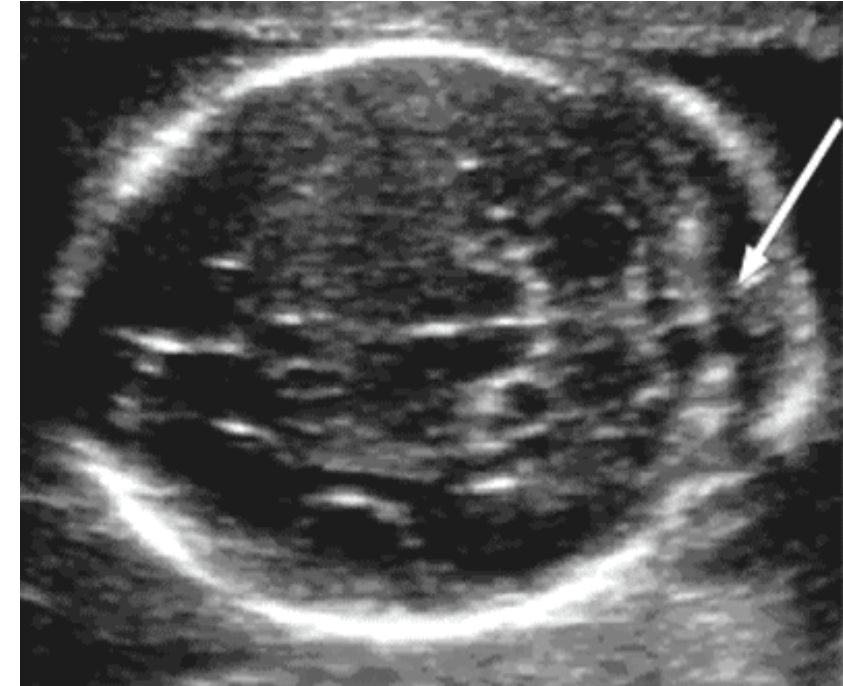
# Ventriculomegaly

- Idiopathic causes
- Chromosomal disorders (the most common is Tri 21)
- Congenital infections (TOXO, CMV)
- Aqueductal stenosis
- Cortical malformations
- Structural abnormalities : corpus callosum agenesis, Dandy-Walker malformation, and neural tube defects, microcephally.



# Increased Nuchal Fold

- The nuchal fold (NF) is the measurement between the outer edge of the occipital bone to the outer margin of the skin in the axial plane of fetal head.
- **NF  $\geq$  6mm** in 16-24 wks of gestation is increased
- **Isolated LR : 3.8** for down syndrome





# Increased Nuchal Fold

- In case of increased NF : Detailed anatomic survey by an expert Genetic counselling Aneuploidy screening vs diagnostic testing even in low risk group

Association with nonchromosomal abnormalities

And Congenital cardiac defects Fetalechocardiography





<b>Finding</b>	<b>Sensitivity Down syndrome percent</b>	<b>False positive rate</b>	<b>Positive likelihood ratio if marker is isolated, percent</b>
Absent or hypoplastic nasal bone	48.9 to 69.9	1.9 to 4	6.58
Hyperechoic bowel	13.4 to 20.7	0.8 to 1.5	5.5 to 6.7
Ventriculomegaly	4.2 to 12.9	0.1 to 0.4	3.8
Increased NF	20.3 to 32.9	0.5 to 1.9	3.79



# pyelectasis

- Pyelectasis is a fluid collection causing dilation of the fetal renal pelvis.
- Renal pelvic imaged in **axial plane** and measure AP diameter:
  - ≥4 mm at 16-27 weeks
  - ≥7 mm at 28-40 weeks

As an **isolated finding**, does not increase the **risk** of aneuploidy.





# Shortened long bones

**Below the 5th percentile for GA** or **Below 2 SD** from the mean

A shortened humerus is a better predictor of DS than a shortened femur (isolated LR of 0.78 and 0.61).

recommend to perform a **detailed ultrasound examination**, determine the etiology and provide counseling

when this is an **isolated finding** and the woman is otherwise at **low risk for fetal aneuploidy**



# Shortened long bones

Mildly short femur may be due to:

Normal variation

Constitutional short limb

False-positive measurement

FGR

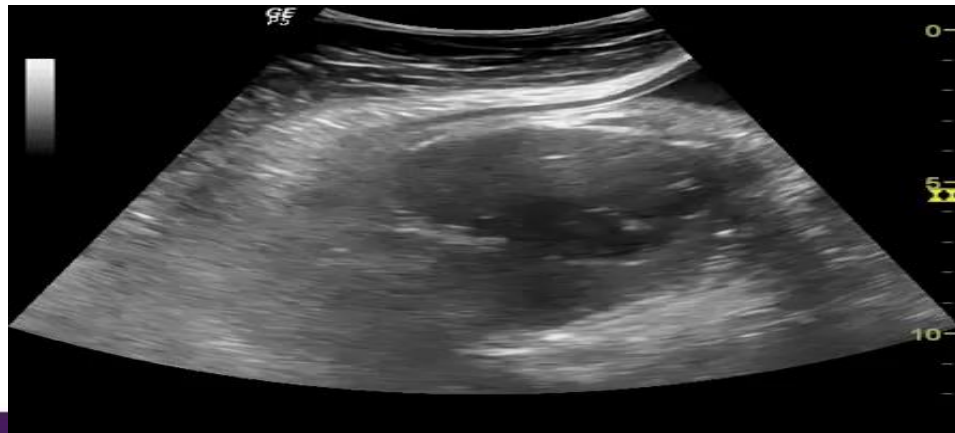
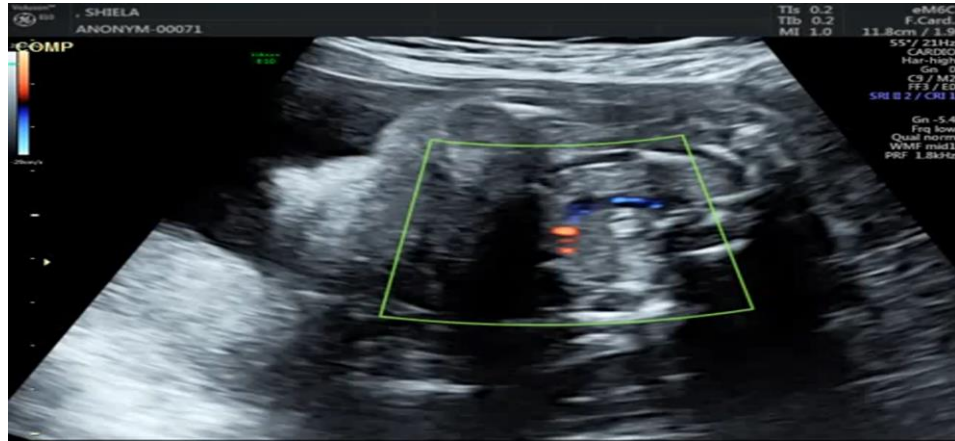
Aneuploidy

Rarely skeletal dysplasia



Online educational program

For more information visit  
[www.fetalmedicine.com/fmf](http://www.fetalmedicine.com/fmf)



## Choroid plexus cyst (CPC) Aberrant Right Subclavian Artery Echogenic cardiac focus

when this is an **isolated finding** and the woman is otherwise at **low risk for fetal aneuploidy**

we recommend to perform a **detailed ultrasound examination and evaluation of heart structure**





Thank you