PITFALLS IN PRE-NATAL DIAGNOSIS OF CONGENITAL HEART DISEASE

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Introduction
Fetal echo has high sensitivity & specificity in prenatal diagnosis of CHD.

However, pitfalls may be encountered, which decrease its accuracy.

We should be aware of these possible pitfalls to **avoid missing important lesions**, & also to avoid **causing unnecessary anxiety** to the parents by a false diagnosis.

What causes pitfalls?

*They are either:*
- due to technical limitations

*Or*
- related to interpretation
What causes pitfalls?

- Fetal
- Maternal
- Or operator /machine limitations

Technical limitations

*May be due to:*

- Poor image quality (unsuitable machine, incorrect settings)
- Fetal position
- Maternal habitus
- Multiple pregnancy
- Oligohydramnios
The most common fetal limitations

- Fetal position (not allowing obtaining views & different planes of imaging)
- Gestational age
- Reduction in amniotic fluid

Maternal: maternal obesity is a common limitation

Pitfalls in interpretation

- Incorrect/false diagnosis (in a normal heart)
- Missed diagnosis (outflow anomalies, minor VSDs)
  
  *(lack of experience & awareness of possible pitfalls)*

- Conditions that develop late in utero
may be erroneously diagnosed as anomalies

- Normal structures in or adjacent to the fetal heart may simulate an abnormality

- Unnecessary referral and concern may be avoided if we are familiar with normal variants

Echogenic Focus in Ventricle

- A small, discrete, echogenic focus in the RV or LV cavity is sometimes seen, especially in the region of the papillary muscles

- Usually of no clinical significance, most likely represents normal papillary muscle or prominent chordae tendineae
Easily distinguished from a cardiac tumor by its typical brightly echogenic appearance, extreme smallness, and location.

Echogenic Focus at Edge of Myocardium

- Occasionally, a brightly echogenic focus at the periphery of the ventricular myocardium (?small tumor, ? myocardial calcification)

- Cause: imaging plane including the anterior end of a rib or part of the sternum.
• Four-chamber view: small echogenic focus at LV apex

• Transducer rotated into a plane that shows a longer section of rib >> pitfall can be detected

Prominent Moderator Band

• MB varies greatly in its development.
• Occasionally apex may be prominent enough to mimic a tumor in RV apex

*Features that favor prominent MB:* singularity & apical location, echogenicity with respect to myocardium, concave edge
Linear Structures Within RA

- Frequently seen in normal fetuses.
- Could be mistaken for abnormally moving flap of the foramen ovale, which is normally seen in LA.

- **Eustachian valve**
- **Chiari’s network**

  (remnants of the embryonic valves of the sinus venosus)

Eustachian valve ("valve of IVC")

- Commonly present

- Appears as a relatively fixed linear echo extending from the junction of IVC with RA toward the foramen ovale

  (slightly oblique 4Ch view)
Chiari’s network

- Infrequently seen
- Several thin, filamentous, mobile echoes in RA
- Move freely at real-time

(The resorptive process of the valve of sinus venosus leaves a fenestrated reticular network of fibers)

Pseudothickening of Tricuspid Valve

- A thickened TV may indicate valve dysplasia.

However, the parietal band ("supraventricular crest"), an arch-shaped band of muscle located between the tricuspid and pulmonary valves, can be mistaken for a thickened tricuspid valve.
Pitfalls in diagnosis of VSD

**Missing or overdiagnosing**

**Pseudo-ventricular septal Defect**

- Occasionally, echoes may be absent in the thinner, membranous portion of IVS, suggesting a VSD
- Typically occurs when the IVS is aligned parallel to the ultrasound beam
How to avoid this pitfall?

- An angle needs to be created between the IVS and the ultrasound beam >> echo drop-out disappears
- If VSD: will persist.
- A true VSD generally has bright margins
- Color flow mapping
Pseudo-apical VSD

False diagnosis of ASD
**Entrance of Coronary Sinus into RA**

- In some imaging planes, the entrance of CS into RA may simulate ASD in the lower part of IAS.

- CS may enlarge in certain forms of anomalous venous return, but also can be identified in many normal fetal hearts.

*Its linear orientation and location help identity it as CS*

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**Pericardial fluid**

- *A small amount can be seen in normal fetal hearts*

- *Normal limit ?? (at least 2 mm)*

*Trace of fluid in normal heart. Note that fluid may not surround entire heart.*
Hypoechoic myocardium

- A related pitfall is the peripheral hypoechoic part of the myocardium, which can mimic pericardial fluid

Can be distinguished from true fluid by observing its continuation into IVS

Pitfalls in examination of outflow tracts

More possibilities of false diagnosis:
Causes of visualization of septo-aortic discontinuity in a normal fetus

- Thinness of the membranous septum
- The path of LVOT courses slightly over the plane of IVS
- Incorrect transducer orientation

How to avoid this pitfall?

- **Careful visualization of LVOT with different insonation angles**
- **Use of color Doppler**
- **Absence of other elements of TOF**
Slightly different imaging plane: overriding is not seen (IVS & aorta are continuous)

Parallel Great Arteries Distal to Valves

- Parallel orientation of aorta and PA at the level of semilunar valves is abnormal and should suggest a transposition abnormality.

- BUT more distally (level of aortic arch) the aorta and pulmonary artery-ductus arteriosus continuum do course parallel for a short segment (may be mistaken for abnormality).
Missing outflow tract anomalies is a serious pitfall as it delays post natal management of potentially fatal conditions

Missing outflow anomalies is more common than septal defects & other lesions detectable in 4 chamber view

TGA is THE MOST COMMONLY missed lesion in fetal echo
TGA: Sequential analysis is mandatory

Tetralogy of Fallot

- Loss of septo-aortic continuity & aortic overriding
- Turbulence at the root of the aorta on color Doppler.
- The RVOT is relatively narrow and reveals poor or no forward flow.
Pitfalls in diagnosis of fetal arrhythmias
Pitfalls in Arrhythmia

- **Technical pitfalls:** difficult to obtain signal
- **Interpretation:** requires thorough knowledge
- **Common mistakes**
  - transient bradycardia
  - intermittent premature contraction

M-mode

- On using M-mode, it is frequently very difficult to obtain a clear signal >>> **pulsed Doppler is preferred**
Frequently
Poor Tracing: difficult to identify wall motion

Occasionally
Good Tracing: Atrial wall contraction precedes mitral valve closure and ventricular contraction
← Ventricular wall
← Aortic valve
← Atrial wall

False diagnosis of bradycardia

• When the fetal heart beat is first visualized at close to 5 weeks of gestation, the rate may sometimes be about 90 BPM, but soon the rate picks up.

• Transient bradycardia is commonly seen at any time (vagal stimulation)
The best time to evaluate the fetal heart is between 18–22 weeks gestation

The fetal heart can be evaluated in the third trimester, **BUT** there are a lot of limitations due to oligohydramnios and shadowing from the fetal spine, ribs, and limbs.
Pitfalls in first trimester evaluation

- Many details are not yet evident
- Some lesions develop later in pregnancy (e.g. outflow obstruction, tumors, cardiomyopathies)
- Adequate examination may be hampered by technical difficulties (image resolution, small size of the heart, fetal movements)

>>>> Cautious reassurance

Lesions with typical late-onset presentation

- Endocardial fibroelastosis
- Cardiac rhabdomyoma
- Hypertrophic cardiomyopathy

- Conditions only diagnosed after birth (PFO, PDA)
Pitfalls in evaluation of chamber size

- Normal size: *Mean ± 2 SD*
- Large heart: Right or left or both sides
- Small chamber: How small?
How small?

- Too small: hardly seen, no potential of growth
- Questionable (CoA, MS, AS ?)
- Not too small: -2 ~ -3 SD

RV dominance in a normal 3rd trimester fetus
How to overcome poor image quality?

- Advanced equipment
- Appropriate adjustment of technical settings (e.g. acoustic focus, frequency selection, signal gain, harmonic imaging, Doppler parameters)
- Longer & repeated examinations
- Experienced sonographer, patience, dedication

Conclusion

- Fetal echo has high sensitivity & specificity in prenatal diagnosis of fetal cardiac anomalies despite some pitfalls
- Overcoming pitfalls by more advanced machines, more skilled operators, adequate timing of study, repeated exams, & good communication with parents can overcome many of these pitfalls & thus lead to better outcome of CHD post-natally