



San Gabriel Valley Perinatal Newsletter

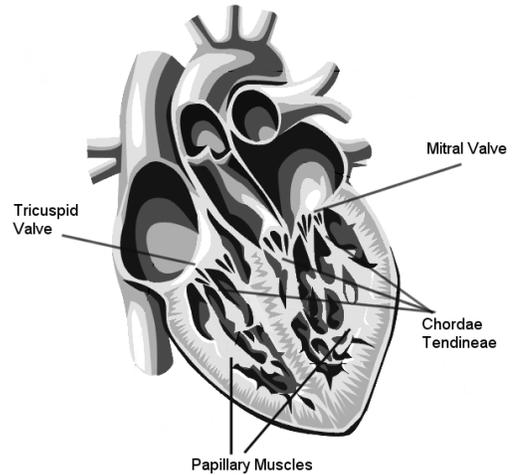
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What is an intracardiac echogenic focus?

The tricuspid and mitral valves of the heart are attached to small papillary muscles within the heart by fibrous strands called the chordae tendineae. The papillary muscles act to stabilize the valves when the ventricles contract.

Echogenic intracardiac foci (EIF) or "bright spots" in the heart are believed to be caused by an increased reflection of sound off the small papillary muscles or chordae tendineae. These bright spots are most often a normal variant and have no significant effect on the development of the heart. EIF may be seen in up to 30, 6, and 11 % of Asian, black, and white fetuses respectively [1]. EIF are found in 13 to 18% of fetuses with Down syndrome [2]. Ultrasound findings that occur more commonly in fetuses with Down syndrome are called markers.



Adapted from Corei Draw 9 Library

The isolated finding of an EIF appears to increase the likelihood of Down syndrome by 1.8 to 2.8 times [3, 4] according to two studies. However, one study found that an isolated EIF in the fetal heart in women aged 18-34 years was not associated with increased risk for Down syndrome [5].

Presently genetic testing of the fetus (amniocentesis) is offered if the risk of Down syndrome in the fetus is 1 in 190 or greater. As the table below illustrates for a woman less than 32 years of age at delivery the finding of an echogenic focus does not increase the likelihood of Down syndrome enough to recommend routine amniocentesis.

Maternal Age At Delivery	Midtrimester Risk of Down Syndrome Based on Maternal Age	Risk of Down Syndrome Based on Maternal Age With EIF
30	1 in 700	1 in 250
31	1 in 613	1 in 219
32	1 in 526	1 in 188

For women 32 years of age at delivery the increased likelihood of Down syndrome, by some reports, would be sufficient enough to offer amniocentesis.

RECOMMENDATIONS

If an echogenic intracardiac focus is found on routine ultrasound then a targeted level II sonogram should be performed

- To distinguish an EIF from a fetal cardiac tumor. Fetal cardiac tumors are most commonly found within the cardiac muscle or septum. Cardiac tumors tend to be larger, multiple, and are not as echogenic as the typical "echogenic focus".
- To identify additional markers for Down syndrome if present.

The use of the EIF as a marker for Down syndrome in the fetus of an Asian mother is questionable due to the increased frequency of EIF as a normal finding in the Asian population.

- In non-Asian populations genetic testing of the fetus (amniocentesis) may be recommended with detection of an isolated EIF, if serum screening (ExAFP) indicates a risk for Down syndrome greater than or equal to 1 in 531 or the patient is 32 years of age or older at delivery.

REFERENCES

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5. Anderson N and Jyoti R Relationship of isolated fetal intracardiac echogenic focus to trisomy 21 at the mid-trimester sonogram in women younger than 35 years. *Ultrasound Obstet Gynecol.* 2003 ;21:354-8.