

# The ultrasound detection of chromosomal anomalies<sup>1</sup>

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## Second trimester findings.

In this section we will review the sonographic markers that can be used in second and third trimester fetuses.

### Cardiovascular anomalies

Cardiac anomalies are very common: 85 per 10,000 newborn will have a cardiac anomaly. But among the fetuses that have aneuploidy, the association is even more dramatic: 99 percent of fetuses with trisomy 18 have cardiac anomaly. Ninety percent of trisomy 13, 50 percent of trisomy 21 etc... therefore there is a great association between the finding of a cardiac anomaly and aneuploidy. Overall 29% of fetuses with a cardiac anomaly also have an aneuploidy, 16% of fetuses that have an isolated cardiac anomaly have an aneuploidy, and when the cardiac anomaly is associated with other anomalies, 66% of these fetuses will have an aneuploidy. Overall the finding of a cardiac anomaly is more related to trisomy 18 and 21 since they are so common, and to a lesser extend to the other aneuploidies.

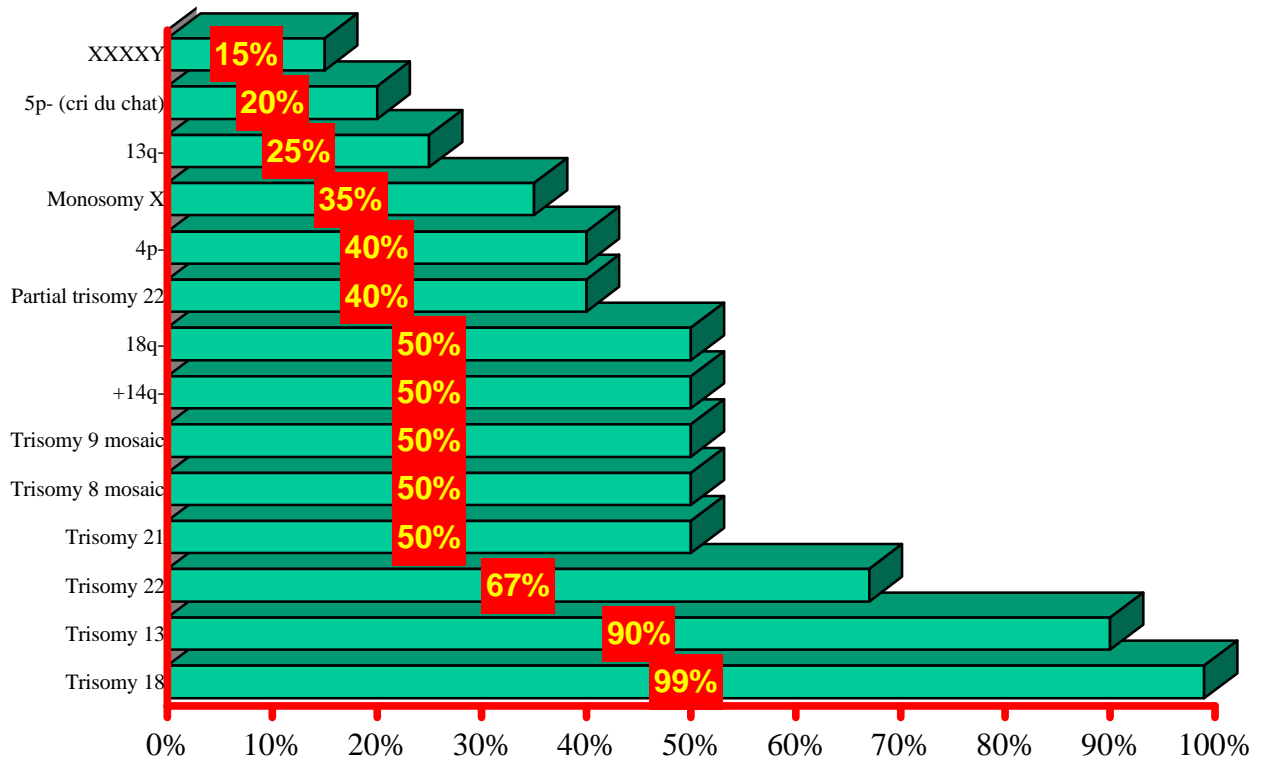


Figure 1: Frequency of cardiac anomalies in several aneuploidies.

### ***Echogenic focus in the heart***

Doug Brown was the first to bring to the attention of the ultrasound community, the association between echogenic foci and trisomy 21<sup>4</sup>. Subsequent papers have demonstrated that about 5% of midtrimester pregnancies have an echogenic foci in the heart. Different authors have reported association between zero and three percent with trisomy 21<sup>5, 6, 7, 8, 9, 10, 11</sup>. The issue of one or more echogenic foci or whether being on the right side or left-sided is more predictive of aneuploidy has not been settled at the current time. The two images demonstrate an echogenic focus in a normal and fetus with trisomy 21: they are no criteria that allows to distinguish the echogenic foci of normal fetuses from those of fetuses with aneuploidies.



*Figure 2 Echogenic focus in the left ventricle of a normal fetus.*



*Figure 3 Echogenic focus in the left ventricle of a trisomy 21 fetus.*

The presence of a small layer of **pericardial fluid** is a common finding. Up to 2 mm is usually considered normal<sup>12</sup> and when it is greater than 2 mm one can think about a pericardial effusion. Pericardial effusions are associated with some aneuploidies and in particular trisomy 21 and to another unrelated topic, which are the TORCH infections.

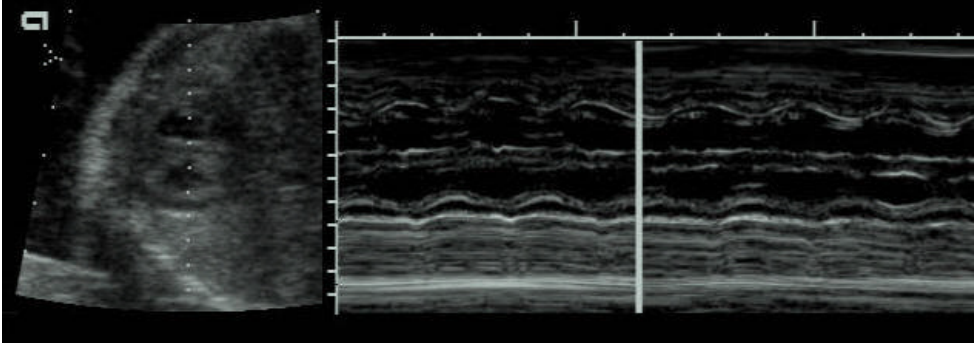


Figure 4: The presence of a small amount of pericardial fluid is often a normal finding, but pericardial fluid may be an indicator of aneuploidy and in particular trisomy 21.

**Hydrops** is a fairly common finding at delivery, 10 per 10,000, and 12-16% of non-immune hydrops can be associated with aneuploidy.

## Chest

Most of the other findings that are present in the chest will be discussed in their respective sections such as the heart, the GI or the skeletal system. About five percent of fetuses with a pleural effusion will have an aneuploidy, and these will mainly be trisomy 21, 13 and monosomy X.

## 2-vessel cord

0.2-1 percent of pregnancies present with a two vessel cord. Among these, about 1-10% have an aneuploidy, including trisomy 18, 13, triploidy and monosomy X<sup>13, 14, 15, 16, 17, 18, 19, 20, 21, 22, 23, 24</sup>.



Figure 5: Sometimes the easiest way to identify a two vessel cord, is not to look at the cord in the amniotic fluid, but to look at the aortic bifurcation. One of the iliac, is much larger than the other iliac, because it carries the blood in the umbilical artery while the other iliac transports blood only for the half pelvis and the other leg.

## Cord cysts

Although most cord cysts are benign finding, occasionally they may be present in fetuses with aneuploidy and in particular with trisomy 13 and trisomy 18.



*Figure 6: Trisomy 13 fetus with a cyst at the base of cord.*

## Placenta

The typical appearance of a Swiss-cheese placenta is that of a placenta that contains innumerable little round vesicles that are quite different from bleed or blood accumulated in the cotyledons. These are much smaller and much more round. The Swiss-cheese placenta is very typical of triploidy but it may also occur in trisomy 18.

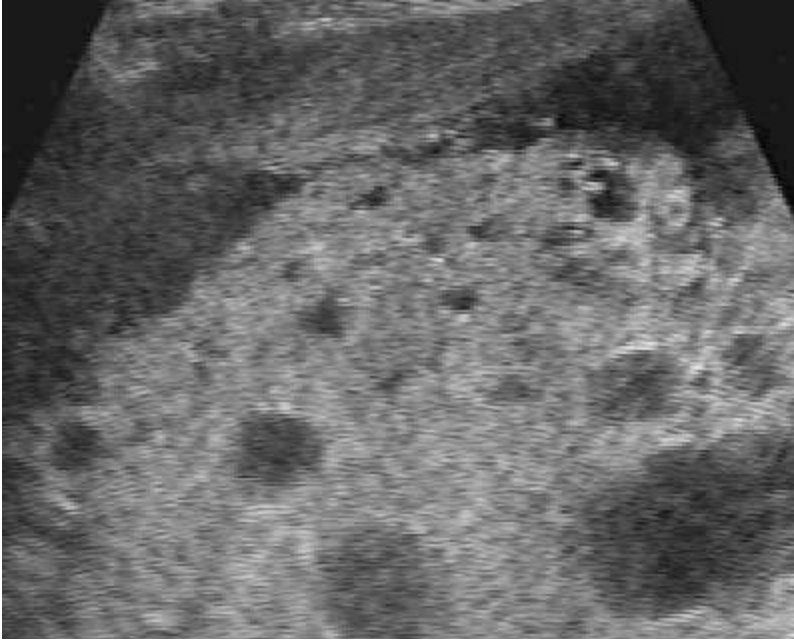


Figure 7: Placental vesicles in triploidy.

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