

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

Werther Adrian Clavelli, MD<sup>2</sup>, Silvia Susana Romaris de Clavelli, MD<sup>2</sup>, Philippe Jeanty, MD, PhD<sup>3</sup>

*Adapted from “The Ultrasound Detection of Chromosomal Anomalies—A multimedia Lecture” by Philippe Jeanty. ISBN (0-9667878-0-3) available at [www.prenataldiagnosis.com](http://www.prenataldiagnosis.com) and [www.TheFetus.net](http://www.TheFetus.net)*

## **Scoring systems**

[Rosaline Schneider and Kipros Nicolaides](#), have developed a very elaborate and well thought scoring system. The system takes into account the age of the patient and the various findings and the risk factor is given to the patient in the form of 1: xxx. The interested reader is referred to their text that contains numerous tables.

The scoring systems that Beryl Benacerraf designed uses minor and major criteria<sup>4, 5, 6</sup>. Major criteria includes major anomalies (such as endocardial cushion defect, omphaloceles and things like that...), a nuchal fold, or a maternal age greater than forty years. The minor criteria include a short femur, a short humerus, pyelectasis greater than 4 mm echogenic bowel, echogenic cardiac focus or a maternal age between 35 and 40 years of age. Major criteria have a score of two and minor criteria a score of 1. In her population a score greater than 2 is associated with a 75 percent risk of trisomy 21, with only a 5 to 10 percent false positive rate. This is a scoring system which is useful because it is simple to use, does not require tables, but it only predicts for trisomy 21.

Our scoring system is even simpler. If the fetus has a major anomalies such as an endocardial cushion defect or omphalocele or any other anomalies that has an intrinsic risk greater than one percent of association with aneuploidy, we offer an amniocentesis. If the fetus has only one small anomaly, such as an echogenic focus in the heart, a choroid plexus cyst, pyelectasis, and the triple screen is normal, and the mother is less than 35 years of age, then we typically do not recommend doing an amniocentesis, unless the patient is very anxious about the finding. Most of these patients do not have amniocentesis. Finally in the occasional fetus that has two small findings such as a single umbilical artery, a choroid plexus cyst, pyelectasis, a simian crease, maternal age greater than 35 years, abnormal triple screen or any of the small findings in combination, we tend to recommend a karyotype because these fetuses are not very common, and the likelihood of aneuploidy is probably greater than one percent in this group too.

## **Differential diagnoses**

A situation that happens from time to time is the finding of a normal amniocentesis in a fetus that has multiple anomalies. There are some syndromes that may resemble aneuploidies such as Smith-Lemli-Optiz, Meckel syndrome, iniencephaly, the cardiosplenic syndromes and the Torch infections. These should occasionally be included in the differential diagnosis of aneuploidies.

## **References**

<sup>1</sup> Adapted from “The Ultrasound Detection of Chromosomal Anomalies—A multimedia Lecture” by Philippe Jeanty. ISBN (0-9667878-0-3) available at [www.prenataldiagnosis.com](http://www.prenataldiagnosis.com) and [www.TheFetus.net](http://www.TheFetus.net)

<sup>2</sup> Diagnostic Maipu, Buenos Aires, Argentina

<sup>3</sup> Women’s Health Alliance, Nashville, TN

<sup>4</sup> Benacerraf BR, Neuberger D, Bromley B, Frigoletto FD Jr Sonographic scoring index for prenatal detection of chromosomal abnormalities. J Ultrasound Med 1992 Sep;11(9):449-58

<sup>5</sup> Nadel AS, Bromley B, Frigoletto FD Jr, Benacerraf BR Can the presumed risk of autosomal trisomy be decreased in fetuses of older women following a normal sonogram? J Ultrasound Med 1995 Apr;14(4):297-302

---

<sup>6</sup> Bromley B, Lieberman E, Benacerraf BR The incorporation of maternal age into the sonographic scoring index for the detection at 14-20 weeks of fetuses with Down's syndrome. *Ultrasound Obstet Gynecol* 1997 Nov;10(5):321-4