



A STUDY OF INCIDENCE OF ECHOGENIC CARDIAC FOCUS IN DIAGNOSING DOWNS SYNDROME:

Radiodiagnosis

Dr Vinod Jacob Associate Professor, Department of Radiology, Travancore Medical College, Kollam

Dr Sonia X James* Professor Department of Radiology, Travancore Medical College, Kollam *Corresponding Author

ABSTRACT

Sophisticated ultrasonography have resulted in the identification of subtle anomalies in the fetus, the significance of which is not always clear. For example, choroid plexus cysts are sometimes seen in the developing cerebral ventricles in mid-trimester. Initially, it was thought that these were invariably associated with the fetus having trisomy 18 but in fact they occur frequently in normal fetuses, although if they are very large and do not disappear spontaneously they can be indicative of a chromosome abnormality. Novel ultrasonographic findings of this kind are often called soft markers, and their interpretation must be approached cautiously in the effort to distinguish normal from abnormal variation. So in cases of Down Syndrome there is high probability of nuchal translucency and other soft tissue markers like Echogenic Cardiac Focus. This study is puts in a sincere effort to find the incidence of presence of EIF in actual confirmed cases of Down syndrome.

KEYWORDS

Introduction:

Ultrasonography offers a valuable means of prenatal diagnosis. It can be used not only for obstetric indications, such as placental localization and the diagnosis of multiple pregnancies, but also for prenatal diagnosis of structural abnormalities not associated with known chromosomal, biochemical or molecular defects.

The frequency of chromosomal anomalies is 1 in 165 [1]. Hook established the relationship between advanced maternal age and fetal aneuploidy [2]. The most common of such markers for aneuploidy include: echogenic intra cardiac focus (EIF). The majority of this associated with an increased risk of trisomy 21 [3], [4]. There is conflicting evidence in the literature regarding not only the management of isolated ultrasound EIF but also its potential association with aneuploidy [5,6]. There is a paucity of information in the literature regarding the prevalence of these markers in different population [7].

Down syndrome patient's most common finding in the newborn period is severe hypotonia. Usually the facial characteristics of upward sloping palpebral fissures, small ears, and protruding tongue prompt rapid suspicion of the diagnosis, although this can be delayed in very small or premature babies. Single palmar creases are found in 50% of children with Down syndrome in contrast to 2% to 3% of the general population. Congenital cardiac abnormalities are present in 40% to 45% of babies with Down syndrome, with the three most common lesions being atrioventricular canal defects, ventricular septal defects, and patent ductus arteriosus.

Novel ultrasonographic findings of this kind are often called soft markers, and their interpretation must be approached cautiously in the effort to distinguish normal from abnormal variation. This study is one such novel effort to find the incidence of presence of EIF in actual confirmed cases of Down syndrome.

Aims and Objectives:

To study and to find the incidence of presence of EIF in actual confirmed cases of Down syndrome.

Materials and Methods:

This study was done in the Department of Radio diagnosis Travancore Medical College, Kollam

The study was done using 60 subjects.

Relevant clinical Examination was done in the Department of OBG, and the scanning was posted in the Department of Radio diagnosis and Imaging.

The patient was asked to drink water and was asked to lie down in a supine position. After applying the USG gel the abdominal scan was

done. The placenta was located and then immediately fetal cardiac shadow was noted. Then the Echogenic Cardiac Focus was tested for hyper echogenicity.

If positivity was found then the other markers were routinely checked for. Confirmed cases of Down syndrome were identified and in them the incidence of Echogenic cardiac focus was confirmed.

Result:

Table 1: Mean age of the study population:

Total	Mean Age	Standard Deviation
30	36.41 years	1.51 years

Table 2: Frequency chart corresponding with age:

Age	18-20 years	20-30 years	30-40 years
Frequency	2	4	54

Table 2: Frequency of Echogenic cardiac Focus Positivity:

Total	Echogenic cardiac Focus Present	Echogenic cardiac Focus Not Found
30	39	21

Table 3: Association of Echogenic cardiac Focus with Downs Syndrome:

Total	X ² value	p value
30	1.62	0.072

So the incidence of Echogenic cardiac Focus is significantly associated with that of Downs Syndrome.

Discussion:

The mean age of the population was found to be 36.41 years and the standard deviation was found to be 1.51 years. The age of the population was on a higher side and is in agreement with the study of Hook EB2. The incidence of Echogenic cardiac Focus is significantly associated with that of Downs Syndrome. The study is in agreement with Andrei Rebarber et al 8. It was found to be significantly associated and it's a powerful tool in India because it is cheap and also forms the first line of diagnosis.

The risk for Down syndrome correlates with absolute values of NT as well as maternal age but, because NT also increases with gestational age, it is more usual now to relate the risk to the percentile value for any given gestational age. In one study, for example, 80% of Down syndrome fetuses had NT above the 95th percentile. By combining information on maternal age with the results of fetal NT thickness measurements, together with maternal serum markers, it is possible to detect more than 80% of fetuses with trisomy 21 if invasive testing is offered to the 5% of pregnant women with the highest. Some babies with Down syndrome have duodenal atresia, which shows up as a

'double bubble sign' on ultrasonography of the fetal abdomen in many centers, it is also standard practice to offer a detailed 'fetal anomaly' scan to all pregnant women at 18 weeks. Although chromosome abnormalities cannot be diagnosed directly, their presence can be suspected by the detection of an abnormality, such as exomphalos or a rocker-bottom foot. A chromosome abnormality is found in 50% of fetuses with exomphalos identified at 18 weeks, and a rocker-bottom foot is a very characteristic, though not specific, finding in babies with trisomy 18, who are invariably growth retarded. The use of other ultrasonographic 'soft markers' in identifying chromosome abnormalities in pregnancy is discussed in the following section. Ultrasonographic 'Soft' Markers sophisticated ultrasonography has resulted in the identification of subtle anomalies in the fetus, the significance of which is not always clear. Novel ultrasonographic findings of this kind are often called soft markers, and their interpretation must be approached cautiously in the effort to distinguish normal from abnormal.

Conclusion:

The incidence of Echogenic cardiac Focus is significantly associated with that of Down Syndrome.

REFERENCES

1. Drugan A, Johnson MP, Evans MI. Ultrasound screening for fetal chromosome anomalies. *American Journal of Medical Genetics*. 2000;90:98-107. doi: 10.1002/(SICI)1096-8628(20000117)90:2<98::AID-AJMG2>3.0.CO;2-H.
2. Hook EB. Chromosome-Abnormalities in Older Women by Maternal Age - Evaluation of Regression-Derived Rates in Chorionic Villus Biopsy Specimens. *American Journal of Medical Genetics*. 1990;35:184-187.
3. Snijders RJM, Shawa L, Nicolaides KH. Fetal Choroid-Plexus Cysts and Trisomy-18 - Assessment of Risk-Based on Ultrasound Findings and Maternal Age. *Prenatal Diagnosis*. 1994;14:1119-1127.
4. Lehman CD, Nyberg DA, Winter TC, Kapur RP, Resta RG, Luthy DA. Trisomy-13-Syndrome - Prenatal Us Findings in A Review of 33 Cases. *Radiology*. 1995;194:217-222.
5. Winter TC, Anderson AM, Cheng EY, Komarniski CA, Souter VL, Uhrich SB, Nyberg DA. Echogenic intracardiac focus in 2nd-trimester fetuses with trisomy 21: Usefulness as a US marker. *Radiology*. 2000;216:450-456.
6. Bettelheim D, Deutinger J, Bernaschek G. The value of echogenic foci ('golfballs') in the fetal heart as a marker of chromosomal abnormalities. *Ultrasound in Obstetrics & Gynecology*. 1999;14:98-100.
7. Shipp TD, Bromley B, Lieberman E, Benacerraf BR. The frequency of the detection of fetal echogenic intracardiac foci with respect to maternal race. *Ultrasound in Obstetrics & Gynecology*. 2000;15:460-462.