

## A STUDY OF INCIDENCE OF ECHOGENIC INTRACARDIAC FOCUS IN SECOND TRIMESTER FOR DIAGNOSING DOWN SYNDROME

*Shashank G. D<sup>1</sup>, Sowmya B. G<sup>2</sup>*

<sup>1</sup>Assistant Professor, Department of Radiodiagnosis, Subbaiah Medical College, Shimoga.

<sup>2</sup>Assistant Professor, Department of Obstetrics and Gynaecology, Subbaiah Medical College, Shimoga.

### ABSTRACT

#### BACKGROUND

Ultrasonography offers a valuable means of prenatal diagnosis. It can be used not only for obstetric indications such as placental localisation and the diagnosis of multiple pregnancies, but also for prenatal diagnosis of structural abnormalities not associated with known chromosomal, biochemical or molecular defects. Ultrasonography is particularly valuable because it is noninvasive and conveys no known risk to the foetus or mother. It does, however, require expensive equipment and a skilled, experienced operator. Sophisticated ultrasonography has resulted in the identification of subtle anomalies in the foetus, 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 foetus having trisomy 18, but in fact they occur frequently in normal foetuses, although if they are very large and do not disappear spontaneously they can be indicative of a chromosome abnormality. Increased echogenicity of the foetal bowel has been reported in association with cystic fibrosis. Initial reports suggested this finding could convey a risk as high as 35%. Earlier it was thought that 10% of the foetuses will have cystic fibrosis, but it is now clear that this risk is probably no greater than 1% to 2%. 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 puts in a hardworking effort 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 Radiodiagnosis, Subbaiah Medical College, Shimoga, Karnataka. This study was done from September 2014 to April 2016. The study was done using 30 subjects.

#### RESULTS

The mean age of the population was found to be 37.28 years and the standard deviation was found to be 1.73 years. The incidence of echogenic intracardiac focus is significantly associated with that of Down Syndrome.

#### CONCLUSION

EIF was found to be significantly associated and can be a powerful tool in India because it is cheap and also forms the first line of diagnosis.

#### KEYWORDS

Echogenic Intracardiac Focus, Down Syndrome, Second Trimester.

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#### BACKGROUND

Ultrasonography offers a valuable means of prenatal diagnosis. It can be used not only for obstetric indications such as placental localisation and the diagnosis of multiple pregnancies, but also for prenatal diagnosis of structural abnormalities not associated with known chromosomal, biochemical or molecular defects. Ultrasonography is particularly valuable because it is noninvasive and conveys no known risk to the foetus or mother. It does, however,

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*Corresponding Author:*

*Dr. Shashank G. D,*

*No. 303, T<sub>3</sub>, Sugandhi Apartments,*

*Gopal Gowda Extension, Shimoga-577205.*

*E-mail: shashankgd@gmail.com, drsowmya.bg@gmail.com*

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require expensive equipment and a skilled and experienced operator. For example, a search can be made for polydactyly as a diagnostic feature of a multiple abnormality syndrome, such as one of the autosomal recessive short-limb polydactyly syndromes that are associated with severe pulmonary hypoplasia- invariably lethal. Similarly, a scan can reveal that the foetus has a small jaw, which can be associated with a posterior cleft palate and other more serious abnormalities in several single-gene syndromes. Until a few years ago, detailed ultrasonography for structural abnormalities was offered only to couples who had a child with a genetic disorder or syndrome for which there was no chromosomal, biochemical, or molecular marker. Increasingly, however, detailed foetal anomaly scanning being offered routinely to all pregnant women at around 18 weeks' gestation as screening for structural abnormalities such as neural tube defects or cardiac anomalies. This



technique can also identify features that suggest the presence of an underlying chromosomal abnormality. Such a finding would lead to an offer of amniocentesis or placental biopsy for definitive chromosome analysis. The future of foetal scanning holds the prospect of three dimensional imaging and magnetic resonance imaging being used more widely and routinely. Although this will clearly enable the unborn baby to be visualised in far greater detail, it will also generate bigger challenges for the radiologist, who might be expected to diagnose serious disorders on the basis of very subtle features.

The frequency of chromosomal anomalies is 1 in 165.<sup>1</sup> Hook established the relationship between advanced maternal age and foetal aneuploidy.<sup>2</sup> The most common of such markers for aneuploidy include- Echogenic Intracardiac Focus (EIF). The majority of this associated with an increased risk of trisomy 21.<sup>3,4</sup> There is conflicting evidence in the literature regarding not only the management of isolated ultrasound EIF but also its potential association with aneuploidy.<sup>5,6</sup> There is a paucity of information in the literature regarding the prevalence of these markers in different population.<sup>7</sup>

Down syndrome (Trisomy 21) is a condition which derives its name from Dr. Langdon Down, who first described it in the Clinical Lecture Reports of the London Hospital in 1866. The chromosomal basis of Down syndrome was not established until 1959 by Lejeune and his colleagues in Paris. The overall birth incidence, when adjusted for the increasingly widespread impact of antenatal screening, is approximately 1:1000 in the United Kingdom, which has a national register. In the United States, the birth incidence has been estimated at approximately 1:1800. In the United Kingdom, approximately 60% of Down syndrome cases are detected prenatally. There is a strong association between the incidence of Down syndrome and advancing maternal age. The 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.

Sophisticated ultrasonography has resulted in the identification of subtle anomalies in the foetus, 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 foetus having trisomy 18 but in fact they occur frequently in normal foetuses, although if they are very large and do not disappear spontaneously, they can be indicative of a chromosome abnormality. Increased echogenicity of the foetal bowel has been reported in association with cystic fibrosis. Initial reports suggested this finding could convey a

risk as high as 35 per thousand. Earlier it was thought that 10% of the foetuses will have cystic fibrosis, but it is now clear that this risk is probably no greater than 1% to 2%. 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 Radiodiagnosis, Subbaiah Medical College, Shimoga, Karnataka.

This study was done from September 2014 to April 2016.

The study was done using 30 subjects.

Relevant clinical examination was done in the Department of OBG, and the scanning was posted in the Department of Radiodiagnosis 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 foetal cardiac shadow was noted. Then the echogenic intracardiac focus was tested for hyperechogenicity.

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 intracardiac focus was confirmed.

**RESULTS**

Total	Mean Age	Standard Deviation
30	37.28 years	1.73 years

**Table 1. Mean Age of the Study Population**

The mean age of the population was found to be 37.28 years and the standard deviation was found to be 1.73 years.

Age	18-20 Years	20-30 Years	30-40 Years
Frequency	Nil	1	29

**Table 2. Frequency Chart Corresponding with Age**

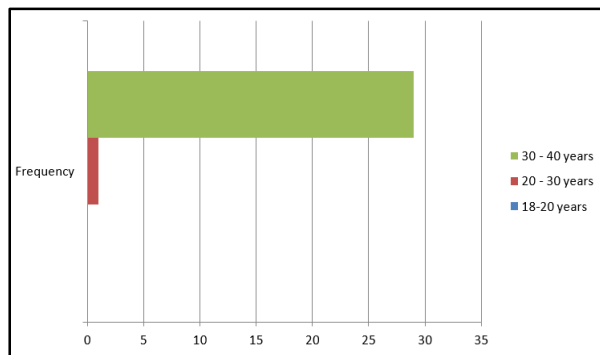
Total	Echogenic Intracardiac Focus Present	Echogenic Intracardiac Focus Not Found
30	18	12

**Table 3. Frequency of Echogenic Cardiac Focus Positivity**

The Echogenic intracardiac focus was found in 18 patients and in 12 patients even though they were confirmed to be Down syndrome no Echogenic intracardiac focus was found.

Total	X <sup>2</sup> value	p value
69	2.34	0.032

**Table 4. Association of Echogenic Intracardiac Focus with Down Syndrome**



**Image 1. Frequency Chart Corresponding with Age**

So, the incidence of Echogenic intracardiac focus is significantly associated with that of Down syndrome.

**DISCUSSION**

The mean age of the population was found to be 37.28 years and the standard deviation was found to be 1.73 years. The age of the population was on a higher side and is in agreement with the study of Hook EB.<sup>2</sup> The incidence of echogenic intracardiac focus is significantly associated with that of Down syndrome. The study is in agreement with Andrei Rebarber et al.<sup>8</sup> It was found to be significantly associated and it is a powerful tool in India because it is cheap and also forms the first line of diagnosis.

Ultrasonography offers a valuable means of prenatal diagnosis. It can be used not only for obstetric indications, such as placental localisation and the diagnosis of multiple pregnancies, but also for prenatal diagnosis of structural abnormalities not associated with known chromosomal, biochemical or molecular defects. Ultrasonography is particularly valuable because it is non-invasive and conveys no known risk to the foetus or mother. It does, however, require expensive equipment and a skilled, experienced operator. For example, a search can be made for polydactyly as a diagnostic feature of a multiple abnormality syndrome, such as one of the autosomal recessive short-limb polydactyly syndromes that are associated with severe pulmonary hypoplasia- invariably lethal. Similarly, a scan can reveal that the foetus has a small jaw, which can be associated with a posterior cleft palate and other more serious abnormalities in several single-gene syndromes

Almost all pregnant women are routinely offered a 'dating' scan at around 12 weeks' gestation. At around this time there is a strong association between chromosome abnormalities and the abnormal accumulation of fluid behind the baby's neck with increased foetal nuchal translucency (NT). This applies to Down syndrome, the other autosomal trisomy syndromes (trisomies 13 and 18), Turner syndrome, and triploidy, as well as a wide range of other foetal abnormalities and rare syndromes. 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 foetal 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 foetal abdomen in many centres, it is also standard practice to offer a detailed 'foetal 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 genetic studies of the parents and the close relatives also has to be studied for understanding the nature and thus the responsibility of the doctors are immense after finding out such findings. The relatives has to be informed and also the fact that consanguineous marriage has its own share of effectiveness in such diseases. The knowledge of such patterns is to be imparted to the patients and thus help them in their desperate time of need. The counseling is an ongoing process and thus is dynamic and not be thought as a sporadic event unless proved otherwise.

**CONCLUSION**

The incidence of echogenic intracardiac focus is significantly associated with that of Down syndrome. It was found to be significantly associated and can be a powerful tool in India, because it is cheap and also forms the first line of diagnosis. The biochemical and genetic tests that has to be done to confirm the diagnosis is FISH and other expensive tests.

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