A Study of Pre-Natal Diagnosis
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**Background:** The genetic diseases that are known to be accompanied with congenital malformations are often not well understood and has an element of surprise attached to it unless proved otherwise as seen in some familial cases. The raw emotions that run in the family of having a new guest, comes to a sudden halt. As the stakes are high and such cases should always be diagnosed as soon as possible a sincere attempt is being made in this study to understand the pre-natal diagnosis using the USG. **Methods:** Nine hundred twenty one patients records of scanning were observed out of which thirty patients who were diagnosed to have some malformations in USG scanning are reported. This study is done in the Department of OBG, Srinivas Institute of Medical Sciences, Mangalore. **Results:** Out of the observed 921 patients thirty was observed to have congenital anomalies. **Conclusion:** USG is able to detect the anomaly and is the gold standard for screening the patients.

**Keywords:** Ultrasound, Non-Invasive, Pre-Natal, Diagnosis, Role.

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**INTRODUCTION**

The genetic diseases that are known to be accompanied with congenital malformations are often not well understood and has an element of surprise attached to it unless proved otherwise as seen in some familial cases. The raw emotions that run in the family of having a new guest, comes to a sudden halt. As the stakes are high and such cases should always be diagnosed as soon as possible a sincere attempt is being made in this study to understand the pre-natal diagnosis using the USG.

**Materials and Methods**

This study was done in the Department of Radiology at Srinivas Institute of Medical Sciences, Mangalore.

Nine hundred twenty one patient’s records of scanning were observed out of which thirty patients who were diagnosed to have some malformations in USG scanning are reported. This study is done in the
Department of OBG, Srinivas Institute of Medical Sciences, Mangalore.

The study was conducted in seventy patients from January to June 2019.

The patients were routinely scanned in the first trimester and then in the second trimester. In the first trimester the Fetal nuchal translucency, the Nasal Bone, Doppler sonographic evaluation of ductus venous blood flow and abnormal tricuspid regurgitation were checked. Enlarged nuchal translucency was noted.

RESULTS

Table 1: First trimester Scan (<2mm Nuchal Translucency)

<table>
<thead>
<tr>
<th>Total</th>
<th>Mean</th>
<th>Standard Deviation</th>
</tr>
</thead>
<tbody>
<tr>
<td>21</td>
<td>1.09</td>
<td>0.14</td>
</tr>
</tbody>
</table>

Table 2: >2 mm Nuchal Translucency (NT)

<table>
<thead>
<tr>
<th>Total</th>
<th>Mean</th>
<th>Standard Deviation</th>
</tr>
</thead>
<tbody>
<tr>
<td>09</td>
<td>2.14</td>
<td>0.23</td>
</tr>
</tbody>
</table>

Table 3: The Nasal Bone (N), Doppler sonographic evaluation of ductus venous blood flow (I) and abnormal tricuspid regurgitation (R)

<table>
<thead>
<tr>
<th>Total</th>
<th>Nasal Bone not developed</th>
<th>Ductus Venous Inverse Flow</th>
<th>Abnormal tricuspid regurgitation</th>
</tr>
</thead>
<tbody>
<tr>
<td>06</td>
<td>02</td>
<td>01</td>
<td>01</td>
</tr>
</tbody>
</table>

Table 4: Other Malformations Found

<p>| | | |</p>
<table>
<thead>
<tr>
<th></th>
<th></th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>Echogenic Intracardiac Focus</td>
<td>11</td>
<td></td>
</tr>
<tr>
<td>Hyperechoic Bowel</td>
<td>02</td>
<td></td>
</tr>
<tr>
<td>Renal Pyelectasis</td>
<td>01</td>
<td></td>
</tr>
<tr>
<td>Choroid Plexus Cysts (CPCS)</td>
<td>03</td>
<td></td>
</tr>
<tr>
<td>Clinodactyly</td>
<td>01</td>
<td></td>
</tr>
</tbody>
</table>

Table 5: Maternal age

<table>
<thead>
<tr>
<th></th>
<th>Without congenital anomalies</th>
<th>With Congenital anomalies</th>
</tr>
</thead>
<tbody>
<tr>
<td>24 ±2.12 Years</td>
<td>34±2.67 years</td>
<td></td>
</tr>
</tbody>
</table>

DISCUSSION

In this study twenty one patients had nuchal translucency less than 2mm with a mean measurement of 1.09mm with standard deviation of 0.14mm. In nine patients the nuchal translucency was more than 2mm with a mean measurement of 2.14mm with standard deviation of 0.23mm. In six patients nasal bone was not developed in 2 patients, ductus venous inverse flow was observed in 1 patient and abnormal tricuspid regurgitation was found in 1 patient. In eleven patients echogenic intracardiac focus was observed, hyperechoic bowel was observed in 02 patients, renal pyelectasis was observed in one patient, CPCS was observed in three patients and clinodactyly was observed in one patient. The mean maternal age was found to be 34 with a standard deviation of 2.67 years further pointing out the fact towards the increased age can be a cause as suggested by other cases conducted by Malone FD et al., [16], and Snijders RJ et al., [17].

None of the Malformations found were interrelated significantly with each other as the test for significance for inter – relation came to be insignificant.

CONCLUSION

The quality of the scan and the ability of the OBG clinician play an important role in diagnosing the fetal malformations in the first trimester. This study may help in the diagnosis at the local level as it tries to give an image of the local incidence of the different malformations.

REFERENCES

8. Falcon 0, Faiola S, Huggon I, et al: Fetal tricuspid regurgitation at the 11 + 0 to 13 + 6-week scan:


