RESEARCH PAPER | MEDICAL SCIENCE | Volume : 6 | Issue : 3 | March 2016 | ISSN - 2249-555X | IF : 3.919 | IC Value : 74.50

Prenatal Sonographic findings of Complete Atrioventricular Septal defect Associated with Arnold – Chiari Type II malformation – A Case Study.

KEYWORDS: Cardiac evaluation, Cardiac abnormalities, Extra cardiac abnormalities, Atrioventricular septal defect, Arnold-Chiari Malformation

Balamurugan P

MBBS,DMRD Research Scholar,Tirunelveli-627002. Tamil Nadu, India (* Corresponding author)

Adaikkappan M

MBBS,DMRD,DNB,PhD.,Professor and Head, Department of Radiology, Rajah Muthiah Medical College, Annamalai university, Annamalai nagar,Chidambaram - 608002, Tamil Nadu,India.

ABSTRACT *Cardiac evaluation during routine second trimester scan, more cardiac abnormalities are detected. Cardiac abnormalities are associated with extra cardiac abnormalities in around 20% in prospectively conducted studies. Presence of combination of cardiac and extra cardiac abnormalities is associated with a risk of a chromosomal or genetic syndrome. Hence, it is important to do a thorough evaluation to look for an extra cardiac abnormality, when a cardiac abnormality is detected. The commonest extra cardiac abnormalities associated with a congenital heart defect include abnormalities of Central Nervous system, Gastrointestinal system, Skeletal and Urinary tract abnormalities. About 24% of the extra cardiac abnormalities are missed on prenatal scan.Here,we present a case of Atrioventricular septal defect(AVSD) with Arnold-Chiari Malformation, which has not been reported before, to our knowledge and as far as our search of literature goes.*

Introduction

Over the years, as the practice of foetal ultrasonography has evolved, a consensus has formed among sonographers, radiologists, obstetricians and maternal-fetal medicine subspecialists: screening for foetal heart disease deserves the dubious distinction of being one of the most challenging and least successful aspects of foetal ultrasonography.(1-3) Because of the challenges inherent with screening the foetus for congenital heart disease (CHD), the rate of prenatal detection of even severe forms of CHD remains disappointingly low.(4-7) Cardiac abnormalities are associated with extra cardiac abnormalities in around 20% in prospectively conducted studies. Presence of combination of cardiac and extra cardiac abnormalities is associated with a risk of a chromosomal or genetic syndrome. The commonest extra cardiac abnormalities associated with a congenital heart defect include abnormalities of Central Nervous system, Gastrointestinal system , skeletal and urinary tract abnormalities.

Atrioventricular septal defects (AVSDs) also known as endocardial cushion defects involve the septal portions of the mitral and tricuspid valves and the adjacent atrial and ventricular septum which is a common cardiac defect in prenatal life.(8)The estimated incidence of the condition varies from 0.33/1000 live births to 0.51/1000 live births.(9)

Complete AVSD (cAVSD) is identified by the echocardiographic hallmark of a common atrioventricular valve and the distortion of the normal appearance at the crux of the heart. Prenatal detection of cAVSD is very important because it is usually associated with chromosomal abnormalities such as Trisomy 21, which occurs in at least 50% of all cases.(10)

Arnold – Chiari Type II Malformation is characterized by small posterior cranial fossa with inferior and downward displacement of cerebellum, fourth ventricle, medulla oblongata , pons (hindbrain structures) below the level of foramen magnum. It is always associated with an open spinal dysrhapism, neuromuscular disorders and skeletal anomalies. Chiari divided these into four types.

Type 1 : herniation of only cerebellar tonsil, not associated with myelomeningocele.

Type 2 :herniation of cerebellar tonsil and brain stem into the spinal canal. Medullary kink, tentorial dysplasia associated with myelomeningocele.

Type 3:associated with cephalocele or cranio cervical meningocele in which cerebellum and brain stem herniated.

Type 4:Associated with marked cerebellar hypo genesis and posterior fossa shrinking.(11) About 24% of the extra cardiac abnormalities are missed on prenatal scan.Here,we present a rare case of combination of Atrioventricular Septal Defect and Arnold-Chiari Type II Malformation with open spina bifida in a fetus by ultrasonography at a gestation of 20-21 weeks (second Trimester) and its management, which has not been reported before.

Case History

A thirty old lady in her first pregnancy was referred for second trimester fetal anomaly scan. She had not undergone previous ultrasound studies, including first trimester NT scan The patient was a nonsmoker, non-alchoholic and had no family history of any genetic disorders. There was no other known medical disorders complicating pregnancy. The weight of the patient was 48 kgs and the height was 5.1 feet.

Ultrasound findings

In fetal face the nasal bone was absent. Premaxillary triangle was seen. In fetal skull, there was inward curvature (scalloping)and deformity of frontal bone, appearances typical of lemon sign. There was mild ventriculomegaly, (width at atrium level was 10 mm), Occipital horns of lateral ventricles were mildly larger than the frontal horns (i.e.) colpo-

Volume : 6 | Issue : 3 | March 2016 | ISSN - 2249-555X | IF : 3.919 | IC Value : 74.50

cephaly small posterior cranial fossa with complete effacement of cistern magna and curving of cerebellum around mid brain, typical of banana sign + . Cisterna magna was absent. Fourth ventricle was not seen.Tentorium cerebelli was pulled downwards. Head circumference was appropriate for the gestational age. There was open spina bifida with a small meningo myelocele seen in the lumbar region. Both feet were normal and there was no evidence of talipes.

In the fetal heart , there was levocardia and normal situs. In four chamber view, there was a large defect seen in the center of the fetal heart. No continuity was seen in between the interatrial septum, inter ventricular septum and the Atrio ventricular valves attachments. The crux was absent. Normal AV valve offsets were not seen. No separate mitral and tricuspid valve components were observed. Both ventricles were of similar size. Aorta and the pulmonary valves were quite similar in size. It was a balanced type of Atrioventricular Septal Defect. There was normal atrioventricular and ventriculoarterial arrangements, no discordance was seen. In Colour Doppler flow, there was communication between all cardiac chambers.Fetal heart rate is 148bpm.It is regular in rhythm.

Discussion:

Complete Atrioventricular Septal Defect is characterized by a large septal defect involving both interatrial and interventricular septa and a common AV valve that connects both atria to ventricles. The affected AV valve is composed of 5 leaflets namely, postero inferior, antero inferior bridging leaflets and three lateral leaflets. Atrioventricular Septal Defect can further be classified as either balanced or unbalanced. In balanced type, the A V junction is connected equally to the right and left ventricle so that each ventricle receives same amount of blood and the ventricles are symmetrical in size. An unbalanced Atrioventricular Septal Defect occurs when the AV junction is predominantly committed to either the right or left ventricle , leading to the hypoplasia of the opposing ventricle receiving the smaller amount of blood. It is associated with other cardiac anomalies like Arnold chiari malformations which is characterized by caudal displacement of hindbrain structures below the level of foramen magnum.

Antenatal diagnosis of complete atrioventricular septal defects is not always easy. When the atrial and septal defects are large, the four-chamber view reveals an obvious deficiency of the central core structures of the heart. Colour Doppler ultrasound can be useful in that it facilitates the visualization of the central opening of the single atrioventricular valve. The atria may be dilated as a consequence of atrioventricular insufficiency. In such cases, colour and pulsed Doppler ultrasound allow the identification of the regurgitant jet.(12,13)

When an AVSD is detected, a complete sequential analysis of the heart is mandatory. Because AVSDs are frequently associated with other cardiac defects. In this study, atrioventricular septal defect was associated with Arnold chiari malformations. Many of the larger studies reported 13–72% association of AVSD with extra cardiac anomalies. (12,13) CHD is responsible for most neonatal mortality but prenatal detection and diagnosis also can improve the outcome of foetuses and neonates with CHD. For these reasons, more improvement needs to be made in prenatal detection of the patient at low risk than in the detailed foetal diagnosis of CHD.

In conclusion, cAVSD is associated with chromosomal, other cardiac and extra cardiac abnormalities. The detection of these abnormalities is important in order to give the best indication of the likely outcome when counselling parents.

Figure -1 Fetal heart showing a central defect in the heart involving the atrial and ventricular septa.

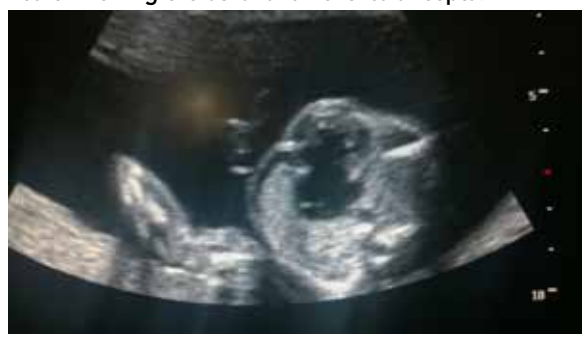

Figure - 2 Fetal heart showing a complete line at atrioventricular junction during systole

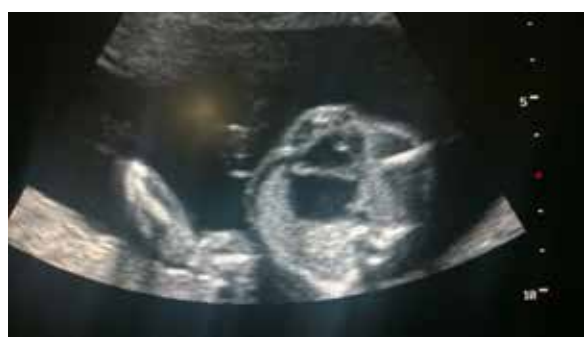

Figure -2 Fetal brain showing ventriculomegaly and a narrow posterior cranial fossa

Figure -2 Fetal brain showing a small meningomyelocele at lumbo sacral region

Volume : 6 | Issue : 3 | March 2016 | ISSN - 2249-555X | IF : 3.919 | IC Value : 74.50

References

1. Allan L, Benacerraf B, Copel J, et al: Isolated major congenital heart disease. Ultrasound Obstet Gynecol.2001;17:370.
2. International Society of Ultrasound in Obstetrics and Gynecology:Cardiac screening examination of the foetus: Guidelines for performing the"basic" and "extended basic" cardiac scan. Ultrasound Obstet Gynecol.2006;27:107.
3. Tegnander E, Eik-Nes S: The examiner's ultrasound experience has a significant impact on the detection rate of congenital heart defects at the second-trimester fetal examination. Ultrasound Obstet Gynecol 2006;28:8.
4. Chaoui R: The four-chamber view: Four reasons why it seems to fail in screening for cardiac abnormalities and suggestions to improve detection rates. Ultrasound Obstet Gynecol.2003.;22:35.
5. Jaeggi E, Sholler G, Jones O, et al: Comparative analysis of pattern, management and outcome or pre- versus postnatally diagnosed major congenital heart disease: A population-based study. Ultrasound Obstet Gynecol.2001;17:380.
6. Garne E, Stoll C, Clementi M; Euroscan Group: Evaluation of prenatal diagnosis of congenital heart diseases by ultrasound: Experience from 20 European registries. Ultrasound Obstet Gynecol.2001;17:386.
7. Heide H, Thomson J, Wharton G, et al: Poor sensitivity of routine foetal anomaly ultrasound screening for antenatal detection of atrioventricular septal defect. Heart. 2004;90:916.
8. Allan LD, Sharland GK,Chita SK,Lockhart S,Maxwell JK.Chromosomal anomalies in foetal congenital heart disease. Ultrasound in Obstetrics and Gynecology.1991;1(1):8–11.
9. Crawford DC , Chita SK , Allan LD : Prenatal defecting of congenital heart disease : Factors affecting obstetric management and survival. Am J Obstet Gynecol 1988 ; 159 : 352 – 356.
10. Marino B.Artioventricular septal defects anatomic characteristic in patients with and without Down syndrome.Cardiol Young 1991 ; 2 : 308 – 310 .
11. Paladini D , Tartaglone A , Agangi A , et al : The association between congenital heart disease and Down syndrome in prenatal life . Ultrasound obstet gynecol 2000; 15 : 104 – 108.
12. Friedberg MK, Kim N, Silverman NH. Atrioventricular septal defect recently diagnosed by foetal echocardiography: echocardiographic features, associated anomalies, and outcomes. Congenital Heart Disease.2007;2(2):.110–114.
13. Huggon IC, Cook AC,Smeeton NC, Magee AG, Sharland GK. Atrioventricular septal defects diagnosed in foetal life: associated cardiac and extra-cardiac abnormalities and outcome. Journal of the American College of Cardiology.2000;36(2)593–601.