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A Rare Case of Omphalocele with Multiple Complex Congenital Anomalies in Intrauterine Fetus

Case Report

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Abstract

In recent times with development of advanced ultrasound equipment and techniques early detection of various fetal anomalies are made easier. Dedicated sonographic technique and interpretation is necessary for early diagnosis of various anomalies. Omphalocele is a type of anterior abdominal wall defect, which is an infrequently reported rare congenital deformity, if detected earlier can help in emergency intervention which can be carried out for a better integrated management for fetus, Mother and family. Special importance needs to be made for Omphalocele as they are associated with multiple other jeopardizing congenital defects leading to poor prognosis. In this case report, we present antenatal imaging findings & gross fetal specimen correlation of fetus of a 21 week primigravida with Omphalocele, Cleft lip and cleft palate, Ebstein's anomaly with hypoplastic left heart, Enlarged posterior fossa of brain with hypoplastic cerebellum and vermis, Large cisterna magna, Hypoplastic orbits and enlarged hyperechoeic kidneys. Of all the investigations, ultrasonographic assessment of fetal structure and well being is most reliable along with other investigations like genetic screening. The lack in the dedicated antenatal examination and poor compliance for routine antenatal checkups leads to a large proportion of undiagnosed cases. Marked reduction in perinatal morbidity and mortality and avoidance of stigma faced by the parents can be achieved by early detection and proper management.

Keywords: Omphalocele; Congenital deformities; Gastroschisis

Introduction

Anterior abdominal wall defects are a group of rare congenital deformities. The incidence of Omphalocele, Gastroschisis, and Exomphalos are worth noting as they frequently turn out to be fatal for the neonates [1]. Special attention should be made Onomphalocele as multiple lethal congenital defects are associated with it which leads to poor prognosis [2].

Omphalocele has been reported to have an incidence of 2-3 in 10,000 pregnancies [3,4]. It is considered to be a defect in regression of the normal physiological umbilical herniation of abdominal content during 6th to 10th week of gestation [4]. The major three diagnostic investigations are targeted fetal anomaly scan for detecting anomalies, echocardiography for detection of various associated lethal cardiac anomalies and lastly karyotyping to detect various chromosomal anomalies which are very often associated with it. Targeted fetal anomaly scan has been the primary imaging modality for the diagnosis and can be used for early identification of the cases by 10th to 12th week of gestation [5].

In developing country like India, where there is lack of perception for routine antenatal checkups among gravid women and lack of proper management of such cases, we have to come up with ideas to increase the efficiency of primary health centres and other health organizations. The strategic measures to be achieved are dedicated ultrasound examination, Proper family history and Routine follow-up examination to assess fetal well-being, awareness programs, and possible genetic screening. A few viable fetuses may be considered, after dedicated assessment, and can be allowed for the continuation of pregnancy with proper surgical and medical follow ups to limit associated complications in such cases. With this marked reduction in perinatal morbidity and mortality can be achieved. We herein report a case of a single live fetus with Omphalocele and multiple associated congenital abnormalities.

Case Report

Written informed consent was taken from the parents of the fetus.

A 20 yr old female of gestational age around 22 weeks came for routine fetal anomaly scan. The anomaly scan showed a single live

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intrauterine fetus corresponding to gestational age 21 weeks 3 days with multiple anomalies as follows: a midline fetal abdominal wall defect is noted at base of umbilical cord insertion with herniation small bowel loops covered with a sac i.e. Omphalocele Fetal echo demonstrates Hypoplastic left atrium and left ventricle, Right atrium is enlarged with apical attachment of tricuspid valve (Atrialization of right ventricle), Single pulmonary trunk is identified from right ventricle i.e. Ebstein's anomaly with hypoplastic left heart. A vertical hypo-echoic region is noted through upper lip in angled coronal view and similar defect is noted in the hard palate in axial plane, i.e. cleft lip and palat. In brain there is enlarged posterior fossa with hypoplastic cerebellum and absent vermis, Note is made of enlarged cisterna magna meas. 13mm. Bilateral kidneys are enlarged and echogenic. Both orbits are small and hypoplastic. No neural tube defects are noted (Figures 1-6).

Post-delivery findings in fetus

Gross fetal specimen with protrusion of small bowel loops covered with membrane noted at umbilical insertion site i.e. Omphalocele (Figure 7(1)). Cleft lip and cleft palate are noted (Figure 7(2)).

Discussion

Improper closure or absence of midline anterior abdominal wall

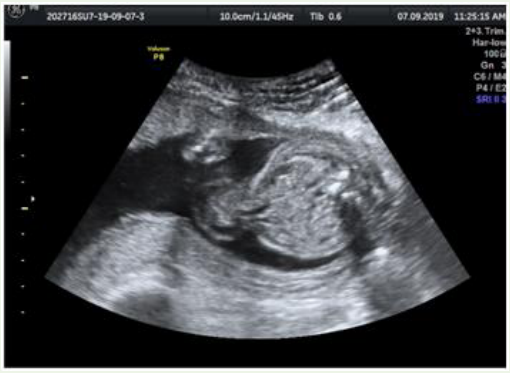

Figure 1: Image showing axial section of 21 weeks fetal abdomen at the level of umbilicus showing herniation of small bowel loops within a sac at level of umbilical cord insertion.

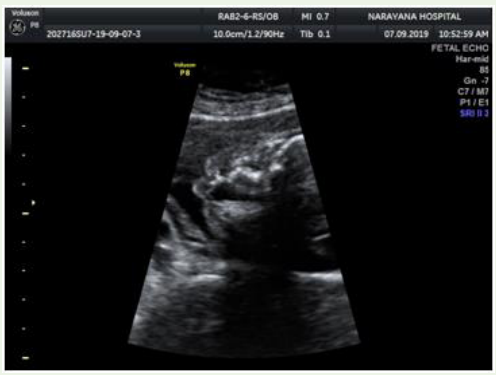

Figure 2: Image showing axial section of 21 weeks fetal face at the level of hard palate showing a vertical hypoechoic line representing cleft palate.

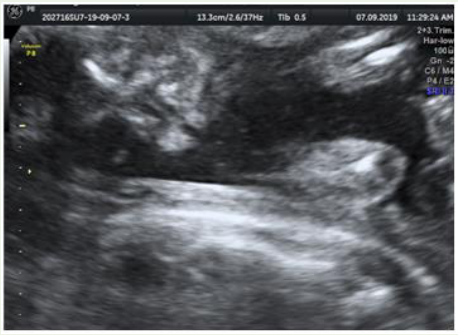

Figure 3: Image showing angled coronal view of 21 weeks of fetal face showing vertical hypoechoic line in upper lip i.e. cleft lip.

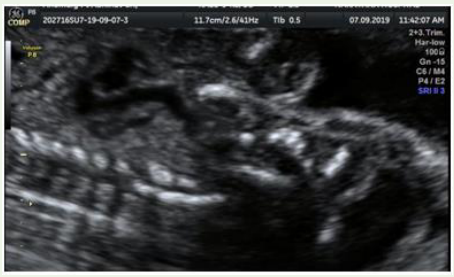

Figure 4: Image showing sagittal section of 21 weeks fetal heart showing hypoplastic left ventricle (1), Single enlarged pulmonary outflow tract (2), Enlarged right atrium, Apical displacement of tricuspid valve with atrialization of right ventricle i.e. features suggestive of Ebstein's anomaly (3).

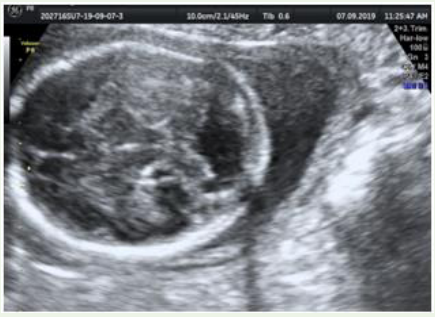

Figure 5: Image showing axial section of 21 weeks fetal brain at level of cerebellum showing enlarged cisterna magna meas.13mm.

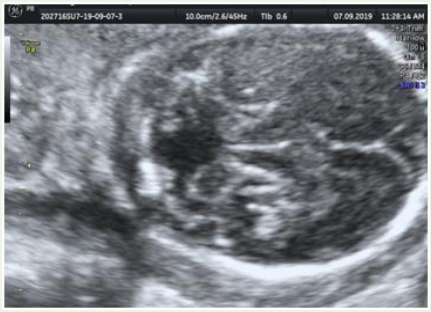

Figure 6: Image showing axial section of 21 weeks fetal brain at level of cerebellum showing absent vermis with hypoplastic cerebellum.

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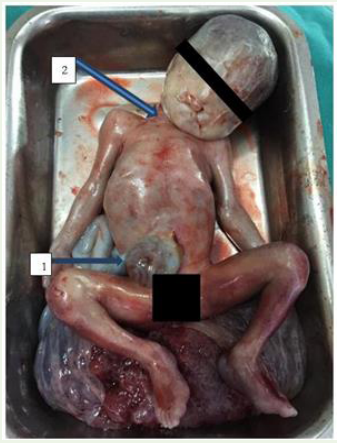

Figure 7: Image showing gross fetal specimen after delivery showing protrusion of small bowel loops covered with membrane at umbilical insertion site i.e. Omphalocele (1), Cleft lip is seen (2).

which may leads to herniation of abdominal contents which results in Omphalocele. According to position of defect they are subdivided into epigastric, Central and infraumbilical omphalocele of which central type is the most common [1]. Studies have showed a strong association of chromosomal abnormalities (69%) with central type of defect of which Trisomy 18 was most frequently noted [2]. Very few patients with such central defect were associated with good prognosis (8%) [1].

Some suggest that Omphalocele may occur due to the failure of the medial segments of the two lateral embryonic wall folds to fuse at approximately 3-4 weeks post conception. This defect at the umbilical ring allows the abdominal content to herniate into a sac comprised of an outer layer of amnion and an inner layer of peritoneum and often Wharton's jelly. General pathogenesis includes teratogenic effects by early pregnancy use of anti thyroid drugs.

Omphalocele (1 in 4000) is rarer than Gastroschisis (1 in 2000) [5]. Etiopathogenesis of Omphalocele and Gastroschisis are debatable and have different theories for origin. Gastroschisis is believed to be formed due to defect in the closure of lateral body wall folds comprised parietal layer of lateral plate mesoderm and overlying ectoderm during the 4th week of gestation [4]. Omphalocele is highly associated with anomalies, and more often small Omphaloceles are more often associated with anomalies. It is associated with multiple chromosomal anomalies like Trisomy 18 (most common), Trisomy 13, Trisomy 21, Turner syndrome, Klinefelter syndrome and Pallister killian syndrome. Other syndrome associations are Beckwith-Wiedemann syndrome, Pentalogy of Cantrell, OEIS complex (Omphalocele, Bladder/cloacal exstrophy, Imperforate anus, spinal anomalies) Lethal Omphalocele- cleft palate syndrome.

After studying 827 cases of Omphalocele from the year 1996 to 2006, Deng et al. established that 52.4% of pregnancies terminated with late fetal death with upward trend in successive years and 37.4% resulted in early neonatal death [2]. The incidence of nonisolated Omphalocele (27.9%) was very less compared to isolated cases (72.1%) evidencing the lesser incidence of syndromic feature of the defect supporting the rarity of our case [2]. The mortality rate for patients in the gestational age of 28-36 weeks was 2.42 times higher than 37-42 weeks and hence earlier diagnosis will give us increased chances toward effective management of more cases well within time [2].

It is associated with various other fetal gastrointestinal anomalies which confer a poor prognosis, fetal anomalies, fetal cardiac anomalies can occur in 50% of cases, Fetal genitourinary anomalies like bladder exstrophy and cloacal exstrophy and fetal skeletal anomalies like Omphalocele-radial ray (ORR) complex.

Higher morbidity and mortality rates are associated with Omphalocele than a Gastroschisis, Primarily due to a higher incidence of associated congenital anomalies. Smaller Omphaloceles are thought to carry a worse prognosis due to increased risk of associated abnormalities.

Mortality rates can approach 80% when associated anomalies are present and increase to ~100% when chromosomal or cardiovascular anomalies exist. However, If found in isolation, then the associated mortality rate decreases to ~10%.

Of all the available investigations, Ultrasonographic assessment of fetal structure and well being is most reliable but the lack in the proper examination and infrequent routine checkups leads to a large proportion of undiagnosed cases. Only about 39.3% of cases are being diagnosed in the health centers and the rest 60.6% are confirmed by physical examination after birth [2]. Proper awareness and proper diagnosis are required to prevent morbidity and social burden on patient's family.

Conclusion

Omphalocele is a rare congenital anterior abdominal wall abnormality associated with multiple congenital anomalies which effects infant mortality and quality of life. Timely diagnosis of this entity and evaluation of other associated anomalies is necessary, which will help in implementation of the optimal treatment protocol and elective termination of the pregnancy. Due to lack of awareness and proper diagnosis, more efficient training of health practitioner, Meticulous reporting, Utilization of modern diagnostic tools such as advanced ultrasound examination and genetic screening should be implemented to reduce the perinatal morbidity and mortality.

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