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PRENATAL SONOGRAPHIC "DOUBLE-BUBBLE" APPEARANCE OF DUODENAL ATRESIA AND ITS ASSOCIATION WITH DOWN SYNDROME

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Abstract:

Objective: *- To describe the prenatal sonographic "double-bubble" appearance of duodenal atresia and further comment on its association with Down syndrome.*

Study Design: *- Single descriptive case study.*

Place: *- Baqai university hospital Township Lahore.*

Materials and Method: *- A 39-year-old patient presented at 26 weeks of gestation for routine obstetric scan. Prenatal sonographic examination was commenced on Toshiba Xario 100 ultrasound machine using a 3-5 MHz curved-array transducer. Trans-abdominal scan was performed. Fetal growth parameters and amniotic fluid volume were measured. Fetal anatomy was assessed for congenital anomalies. Amniotic fluid cells sample was obtained to carry out prenatal cytogenetic analysis.*

Results: *- Antenatal ultrasound scan revealed a single intrauterine active male fetus with over- distended stomach and duodenum giving rise to a "double-bubble sign" and associated mild polyhydramnios was noted indicating duodenal atresia. Suspicion of Down syndrome was raised. Prenatal cytogenetic investigation (amniocentesis) was carried out to rule out any presence of chromosomal anomalies. The test confirmed that the fetus had Down syndrome. There was confirmed association of duodenal atresia and Down syndrome in the fetus. Family decided to continue pregnancy which was managed accordingly and carefully planned intervention resulted in a live-birth. Duodenal atresia was treated surgically to improve survival rate. This immediate intervention proved beneficial.*

Conclusion: *- Ultrasonography is a safe modality for prenatal screening. There is a known association between duodenal atresia and Down syndrome (approximately 30%) which was noted in our study. Duodenal atresia reveals a typical "double-bubble sign" on prenatal ultrasonography. Awareness of this prenatal sonographic finding should result in improved detection of Down syndrome and also prompt a cytogenetic investigation which will aid to identify, plan and direct the management of fetuses at risk of Down's syndrome.*

KEYWORDS: *- Ultrasonography, Duodenal Atresia, Down Syndrome, Double-Bubble Sign*

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INTRODUCTION

Duodenal atresia is a congenital fetal gastrointestinal structural anomaly; its occurrence is approximately 1: 10,000 live births[1]. Duodenal obstruction can be classified as either intrinsic (duodenal atresia, stenosis. webs) or extrinsic (annular pancreas, Malrotation)[2]. Duodenal atresia is an intrinsic type of duodenal obstruction[3].Atresia refers to congenital absence[3]. Duodenal atresia causes complete obstruction of the duodenum[4]. It occurs due failure of the duodenal lumen to recanalize during 8 to 10 weeks of gestation[4].In 80% of the cases, the obstruction is usually distal in the second portion of duodenum at the ampulla of vater while 20% obstructions are proximal[5]. Anatomically, duodenal atresia can be classified into three types Type 1 (92%) where there is complete obstruction with intact membrane, Type 2 (1%) atresia connected with a fibrous cord separating two ends, Type 3 (7%) atresia with blind-end and mesenteric separation[5].Duodenal atresia is associated with multiple anomalies namely Down's syndrome, annular pancreas, Meckel's diverticulum, cardiac malformations and malrotation[5]. Approximately 30% of duodenal atresia cases are associated with Down's syndrome[5]. Incidence of Down syndrome is 1/700 live births[6]. Mostly Down syndrome is due to maternal non-disjunction during meiotic division that results in an extra chromosome 21 (trisomy 21).[6]In 3-4% Down syndrome is due to translocation with normal chromosome number but with additional chromosome 21 linked with chromosome 14[6].Rarely Down syndrome is due to mosaicism with some cells having normal chromosomes while other cells having abnormal 47 chromosomes[6].Risk factors for Down syndrome include advanced maternal age, family history of chromosomal abnormalities, previous child with chromosomal abnormality, X-ray exposure, abnormal maternal serum marker value[7]. The prevalence of duodenal atresia in Down syndrome fetuses is approximately 3%[4].

CASE STUDY:

A 39-year-old women, gravida 2, and at 26 weeks of gestation, was referred to Baqai university hospital in Township Lahore for third trimester routine scan. The patient was neither diabetic nor hypertensive and overall reported no history of medical illness till date. There was no family history of chromosomal abnormalities and pregnancy histories as well as previous obstetric scans were unremarkable. Prenatal sonographic examination was commenced on Toshiba Xario 100 ultrasound machine using a 3-5 MHz curved-array transducer. Trans-abdominal scan was performed. Scan revealed a single-ton male intra-uterine gestation. The fetus was active with cephalic presentation and longitudinal lie. Regular fetal heart beat and growth was noted. Scan revealed a gastric "double-bubble sign" in the upper part of fetal abdomen corresponding to over-distended fetal stomach and duodenal bulb, indicative of fetal duodenal atresia causing duodenal obstruction. Mild polyhydramnios was noted (deepest vertical pocket measuring 8cm). No other fetal anomalies were detected.

The abnormal sonographic findings raised suspicion for likely diagnosis of Down's syndrome. The family was counselled for amniocentesis to confirm diagnosis. Chromosomal amniotic fluid cells analysis confirmed the diagnosis of Down's syndrome revealing a 47 XY, +21 chromosomes. Family decided to continue pregnancy. Pregnancy was monitored timely. Fetus did not develop any additional congenital anomalies throughout the remaining gestation. Delivery was managed accordingly and with thoroughly planned interventions live-birth resulted. Post-natal evaluation confirmed diagnosis of duodenal atresia. Duodeno-duodenostomy was performed to improve survival rate. Post-operative recovery was eventful.

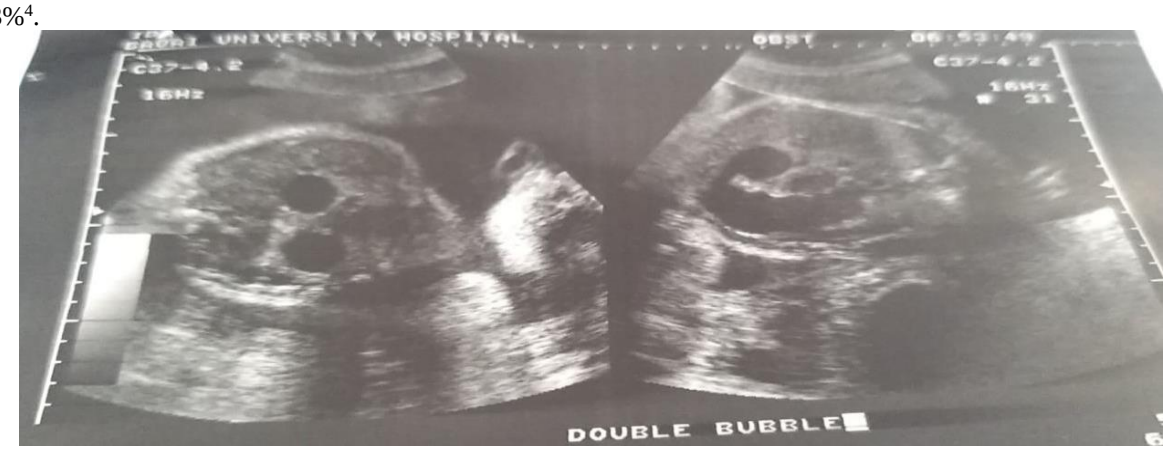

Figure 1: - Duodenal atresia: - **LEFT IMAGE –** Axial image of the fetal abdomen at 26 weeks shows the double-bubble sign of over-distended stomach in the left upper quadrant and over-distended duodenal bulb to the right of the midline. **RIGHT IMAGE –** after adjusting the scan plane to assess for communication between the two fluid-filled structures confirms a connection.

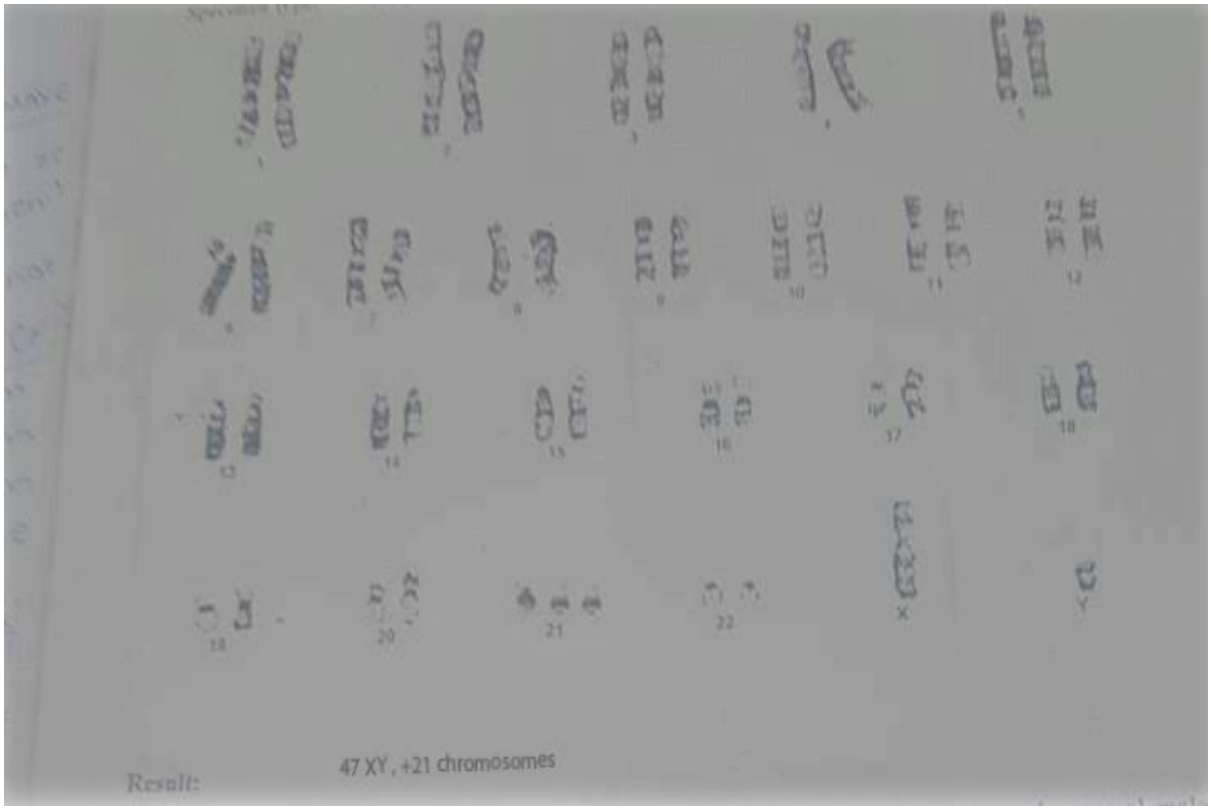

Figure 2: - Third trimester cytogenetic amniocentesis revealed 47 XY, +21 chromosomes confirming that the male fetus has Down syndrome.

DISCUSSION:

Duodenal atresia is complete obstruction of the proximal duodenal segment causing abnormal dilatation of fetal duodenal bulb and stomach as seen on the present prenatal sonogram. Duodenal obstruction can be classified as either intrinsic or extrinsic[3].The present fetus had duodenal atresia which is an intrinsic type of duodenal obstruction. Duodenal atresia can be diagnosed effectively after 22-24 weeks of gestation[9]. Diagnosis in early pregnancy is difficult due to immature fetal gastro-intestinal tract emptying[9]. In current study, duodenal atresia was clearly visualized at 26 weeks of gestation.

Bishop J.C Et al retrospective cohort study (2008-2017) concluded that the "double-bubble sign" on antenatal sonography is a reliable indicator of duodenal atresia[11]. Analysis of prenatal and postnatal data revealed that all the live-births who showed prenatal double-bubble sonographic appearance had duodenal atresia confirmed on post-natal evaluation thus providing a positive-predictive value to be 100%. This sign is the hallmark of the current study in which the "double-bubble" signifies presence of two fluids -filled structures visualized in the upper portion of fetal abdomen on the transverse plane. Residing in the upper left quadrant of fetal abdomen is a large anechoic dilated cystic structure (first bubble) which is the distented fetal stomach. On its right, a smaller anechoic cystic structure (second bubble) represented the distended fetal duodenal bulb. These two structures were separated by a gastric antrum (hypoechoic). Distal to the site of obstruction, no gas was present. To rule out false-positive results, the diagnosis of duodenal atresia was manifested only when the two-fluid filled structures were intercommunicating on longitudinal scan plane (a diagnostic key). Duodenal atresia associated polyhydramnios is seen (50%) of the cases particularly in the third trimester owing to inability of fetus to swallow fluid and blocked passage of fluid[10].Mild polyhydramnios was noted in the current study as well thereby strengthening the diagnosis. Once duodenal atresia is positively diagnosed, it is important to search for associated congenital fetal anomlies[10]. Bishop Et al study further conducted additional post-natal cytogenetic, molecular and clinical genetic analysis of the subjects which revealed that 21 cases of duodenal atresia in the study had associated genetic abnormalities of which Down syndrome was confirmed in 6 cases[11]. In the present case study, down syndrome presence was confirmed in fetus diagnosed with duodenal atresia prenatally.

Sonographic differential diagnosis of duodenal atresia documented in C.M Badiu Et al study in 2019 need to be ruled out in order to eliminate false-positive diagnosis[4].Some other etiologies of fetal intestinal obstruction can simulate the double-bubble sign on the sonogram[4].Annular pancreas is

the second most common cause of duodenal obstruction. Contrary to duodenal atresia, annular pancreas is an extrinsic type of duodenal obstruction. It causes partial duodenal obstruction. On prenatal sonographic scans, presence of annular pancreas can be ruled out on basis of visualization of an additional hyperechogenic band circling the duodenum[4]. This is not seen in case of duodenal atresia. Jejunal obstructions manifest much more distally and often more than two cystic bubbles on fetal sonogram may be seen[4].Duodenal stenosis is the narrowing of the intestinal lumen which produces much less dilatation of fetal stomach and duodenum and the associated double-bubble produced is much smaller in size and less prominent than that seen in duodenal atresia and additionally gas distal to the site of narrowing may be seen[10].In fetus with anomalous bile duct anatomy, presence of distal bowel gas is noted which needs to be ruled out for accurate diagnosis of duodenal atresia[4]. This mentioned list of differential diagnosis of fetal duodenal atresia was ruled out carefully in the current study to avoid any false-positive diagnosis.
Ultrasonography proves to be valuable for earlier diagnosis of duodenal atresia (during the antenatal period) based on the double-bubble sign[8]. Delayed diagnosis and presentation constitutes one of the main contributing factors for increased mortality rate in patients having duodenal atresia owing to complications such as electrolyte disturbance, aspiration, dehydration and pnemonia[10]. Bilal mirza Et al study in 2012 reported three cases of neonatal duodenal atresia diagnosed on post-natal evaluation and assessed them for presence of any other associated anomalies[5]. All the three subjects had associated Down syndrome confirmed on post-natal karyotyping. Unfortunately, two of the subjects expired owing to delayed post-natal diagnosis and other presence of additional anomalies[5]. Comparatively, in the current study, diagnosis of duodenal atresia and associated Down syndrome was documented much earlier i.e during the prenatal period so that the prompt planning options and therapeutic interventions that followed resulted in live-birth and further immediate interventions allowed for eventful recovery of the subject.

Duodenal atresia accounts for one-third of fetuses with Down's syndrome[4]. S.Yagel El al proposed fetal duodenal atresia to be a high risk indicator for Down's syndrome[12].The association is confirmed in this case study.Although cytogenetic amniocentesis is a reliable diagnostic intervention for chromosomal abnormalities in-utero, it is invasive and is associated with increased risk of membrane rupture[7].Ultrasound is a safe modality for prenatal screening[4].This highlights the need for awareness of prenatal sonographic associations of Down syndrome to be documented so that cytogenetic amniocentesis can only be performed in specific cases.In present case, cytogenetic amniocentesis was only carried out to confirm Down syndrome in fetus which was suspected when antenatal sonogram revealed the double-bubble sign typical of duodenal atresia.

CONCLUSION:

Duodenal atresia is a high risk indicator for Down syndrome. There is an approximate 30% association of duodenal atresia and fetuses with Down syndrome. This rare association was confirmed in the present case. Ultrasonography is the only safe imaging modality for prenatal screening. Awareness of the aforementioned prenatal sonographic sign "double-bubble sign" which is typical of duodenal atresia proves beneficial to identify, plan and direct the management of fetuses at risk of Down's syndrome.

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