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Antenatal Diagnosis of a Rare Fetal Syndrome: Is This a New Syndrome or a Variant of Goldston Syndrome?

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Goldston Syndrome is a rare condition characterized by polycystic enlarged kidneys and Dandy-Walker Malformation. It is a rare variant of Meckel Gruber Syndrome.

A case with a possible diagnosis of Goldston syndrome is presented; the diagnosis was made by fetal ultrasound at 30 weeks of gestation in 25 years old Saudi mother who had previous pregnancies with congenital malformations. Ultrasound of the fetus showed severe olighydramnios, Dandy-Walker malformation, enlarged echogenic kidneys and fetal ascites. To my knowledge, this is the fifth case of Goldston syndrome diagnosed during intrauterine life or could be a new entity because of fetal ascites.

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Goldston Syndrome (GS) is rare entity, characterized by renal dysplasia and Dandy-Walker Malformation (DWM)[1-3]. The most common syndrome associated with renal and central nervous systems abnormalities is Meckel Gruber syndrome. Goldstone syndrome considered a variant of Meckel Gruber syndrome[4]. Antenatal ultrasound is an important tool for fetal diagnosis. The combination of fetal enlarged echogenic kidneys, Dandy-Walker malformation with olighydramnios is suggestive of Goldston syndrome, in our case there was an extra ultrasound finding of fetal ascites.

The aim of this report is to present a rare case of Goldston syndrome with fetal ascites.

THE CASE

A twenty-five years old Saudi, she was Gravida 8, Para 7, having living three only. She was referred for further management because of suspected fetal anomalies.

Her past obstetric history is significant. Her first baby was a boy 10 years old now and healthy, delivered vaginally at term. The second pregnancy was intrauterine fetal death of girl at 9 months delivered by cesarean section for unknown reason.

The third pregnancy was a boy 7 years old now and healthy, delivered vaginally at term. The fourth pregnancy was a girl 5 years old now and healthy, delivered vaginally at term. The fifth was a boy, the sixth was a girl, the seventh was a boy, all of them were intrauterine fetal deaths at 8 months of pregnancy and all were vaginal delivery.

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The wife and husband are cousins, the wife is illiterate and the husband is soldier with primary school level.

Ultrasound at 30 weeks of gestation showed single viable fetus, biometry at 30 weeks, but with severe oligohydramnios. Fetal brain showed Dandy-Walker malformation and very small chest compared to the fetal abdomen, see figures 1 and 2. The heart could not be well seen. Fetal abdomen showed very huge hyperechoic kidneys (solid in texture not cystic) almost occupying the whole abdomen, also there was a significant fetal ascites, fetal bladder could not be seen, see figures 3 and 4. Spine and limbs looked normal.

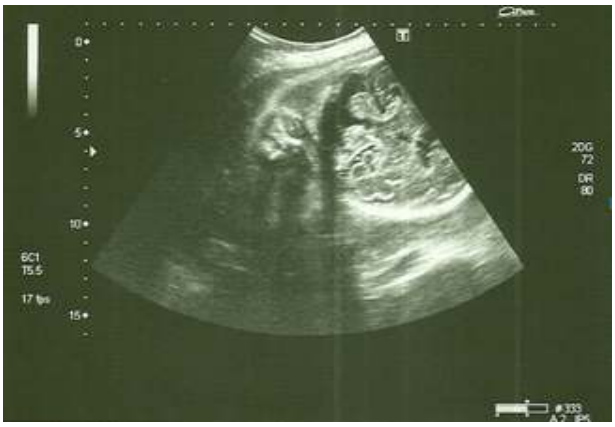

Figure 1: Fetal Ultrasound of the Brain Showing Dandy-Walker Malformation

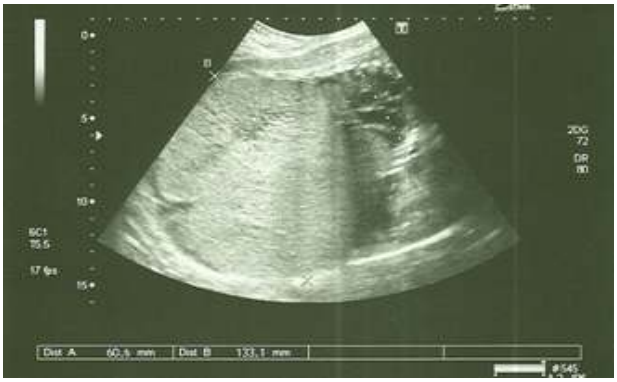

Figure 2: Fetal Ultrasound of the Fetal Chest and Abdomen. The Diameter of Fetal Abdomen Is Enlarged, Compared with Very Small Chest Diameter

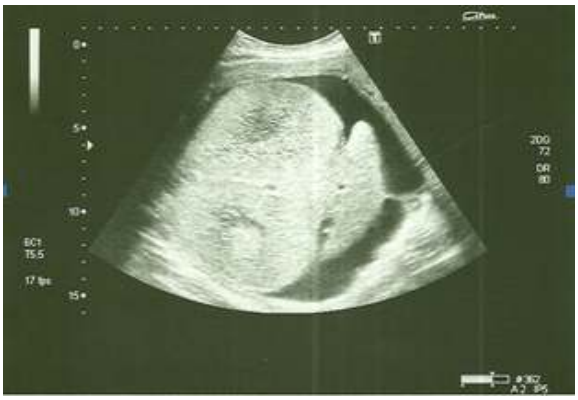

Figure 3: Fetal Ultrasound of the Abdomen Showing Enlarged Symmetrical Echogenic Kidneys with Fetal Ascites (Liver Seen on Top of the Kidneys Floating in the Ascetic Fluids)

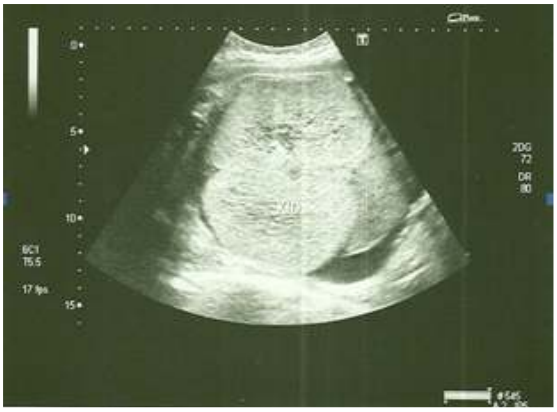

Figure 4: Another View Showing Very Large Echogenic Kidneys Occupying Almost the Whole Fetal Abdomen

At 36 weeks of gestation, she presented to emergency room in active labor and had emergency cesarean section due to breech presentation. The outcome was a girl with a birth weight 3 kilogram. APGAR score was 0, 1, 2, at 1, 5, 10 minutes respectively.

The baby died the next day in neonatal intensive care unit. The family refused autopsy for religious reasons, also refused any referral for both of them for further genetic testing and counseling.

DISCUSSION

Association of polycystic kidneys with Dandy-Walker malformation (DWM) has been referred to as Goldston syndrome (GS) or cerebro-renal syndrome, which is a rare familial disorder[1,3]. The kidneys are cystic and enlarged bilaterally. The brain shows DWM, which is the cystic dilatation of the fourth ventricle secondary to obstruction of foramina of Luschka and Magendie, agenesis or hypoplasia of the cerebellar vermis and enlargement of the posterior fossa[4]. Hepatic dysplasia/fibrosis and pancreatic dysplasia are other disorders that could be seen in GS[1,5].

The fetus presented had very large hyperechoic kidneys. Chaumoitre et al described eight different genetic syndromes in 93 fetuses presenting with hyperechogenic[6]. One-third of those fetuses had renal cysts with other associated malformations, which were more useful in the diagnosis than the renal cyst. If no malformation is found, the main diagnosis remains polycystic kidney disease.

Two of the three major anomalies (bilateral polycystic kidneys, occipital encephalocele and post-axial polydactyl) are diagnostic signs of Meckel-Gruber syndrome, which is a lethal autosomal recessive disorder. It was reported that 57% of the cases had three cardinal findings and 16% had only polycystic kidneys with polydactyly[7].

Dandy-Walker malformation and cystic dysplastic kidneys seen in Goldston syndrome could be seen in MGS. Differentiation between these two syndromes is possible by the fact that polydactyly and occipital encephalocele are absent in GS. However, it has been debated that these two entities are similar and GS has been accepted as a variant of MGS[8-10].

Genetic characteristic of this fatal syndrome has been described as autosomal recessive, as in Meckel-Gruber syndrome[2,11,12].

Gloeb et al described the first case of GS in the seventeenth week of gestation. The condition was diagnosed by US in 635 gm fetus. They suggested Goldstone might be considered as a part of the spectrum of Meckel-Gruber syndrome[13]. Gulcan et al described the second case of Goldston syndrome in the twenty-eighth week of gestation, which had Dandy-Walker malformation, cystic dysplastic kidneys, structural cerebellar anomalies and other associated anomalies (large fontanel, low set ear, hypertelorism, retromicrognathia)[1].

The third reported case of Goldston syndrome in intrauterine life was described by Avcu et al, where the obstetric ultrasound and fetal MRI studies showed hydrocephalus, agenesis of the cerebellar hemispheres, vermian hypoplasia, cystic dilatation of the 4th ventricle, enlargement of the posterior fossa, abdominal distension and oligohydramnios. The kidneys were symmetrically enlarged and multicystic in a fetus of twenty-second week of gestation[14].

The fourth case of Goldston syndrome in intrauterine life was described by Hussain et al, where the ultrasound showed a single fetus corresponding to 27 week of gestation and ultrasound picture of Dandy-Walker malformation. The kidneys were enlarged and echogenic containing small cysts with associated oligohydramnios[15].

To the best of my knowledge, this case is the fifth case of possible Goldston syndrome diagnosed during intrauterine life. Due to religious and social believes we could not do autopsy studies on the fetus, also the parents refused to go for further genetic testing. The only difference in this case is the presence of huge kidneys, which are occupying the whole abdominal cavity and significant ascites; these two findings leading to a question: Are we dealing with a variant of Goldston syndrome or is this the first reported case of new genetic syndrome?

CONCLUSION

A rare case of Goldston syndrome with ascites was presented. The baby was delivered by cesarean at 36 weeks of gestation. The baby died the next day, in the neonatal Intensive Care Unit.

Potential conflicts of interest: No

Competing interest: None **Sponsorship:** None

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