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Gastroschisis Coexistent with Turner Syndrome: A Case Report

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Gastroschisis is a rare anomaly and it is usually not associated with other syndromic and nonsyndromic anomalies. This is the case of gastroschisis with aneulpoidy (Turner syndrome). A fetal huge cystic hygroma was diagnosed by prenatal sonography at 14 weeks' gestation. Combined hydrothorax was diagnosed at the same time. The pregnancy was terminated by induction of labor at 15 weeks' gestation. The female fetus had a big membrane of cystic hygroma surrounding the fetal neck with multiple septations. Additionally, a full abdominal thickness defect with multiple loops of bowel outside the abdomen, which could not be diagnosed on prenatal ultrasound scan, was detected on postnatal examination. It would be possible support explanations for the co-existence of gastroschisis with cystic hygroma might be compression of the cystic mass on the abdominal wall causing occlusion of the terminal segment of the right omphalomesenteric artery or the flow-related anomalies of the cardiovascular system commonly seen in Turner syndrome.

Key words: Gastroschisis, Turner syndrome, Fetal anomaly

서 론

Gastroschisis is a birth defect characterized by a usually small and right-sided abdominal wall defect, through which abdominal organs, mainly the midgut, protrude. Its etiology and pathogenesis are unknown, although vascular occlusion, possibly of the omphalomesenteric artery, has been considered as probably involved.[1,2] The incidence of gastroschisis has been reported to be 1.36 in 10,000 live and stillbirths.[3] Furthermore, young maternal age and rising secular trends are two outstanding epidemiologic characteristics of gastroschisis,[4] the meaning of which is unclear. It has been noted that gastroschisis neither has an increased risk of chromosomal abnormalities nor has it been associated with other extra-gastrointestinal anomalies.[5]

Turner syndrome (or Ullrich-Turner syndrome) is the most common sex chromosome abnormality in female fetuses and is caused by a complete or partial X monosomy in some or all cells. Typical sonographic findings of Turner syndrome are nuchal cystic hygroma (CH), nonimmune hydrops, and renal and cardiac defects6 however, neither sonography nor maternal serum screening should be considered diagnostic of Turner syndrome, and karyotype confirmation (either by amniocentesis [AC] or by chorionic villus sampling [CVS]) should be obligatory.[7]

Although gastroschisis tends to be an isolated anomalynot associated with Turner syndrome, we report a case of the literature of gastroschisis co-existing with Turner syndrome.

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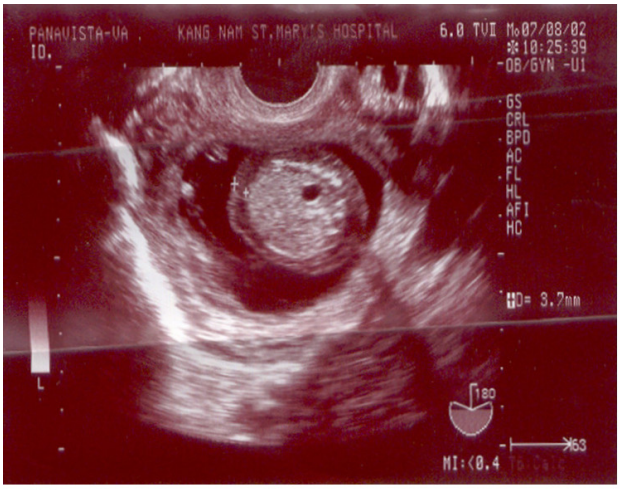

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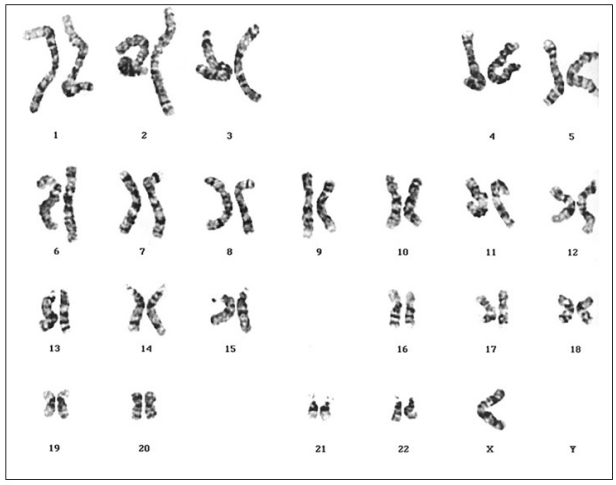

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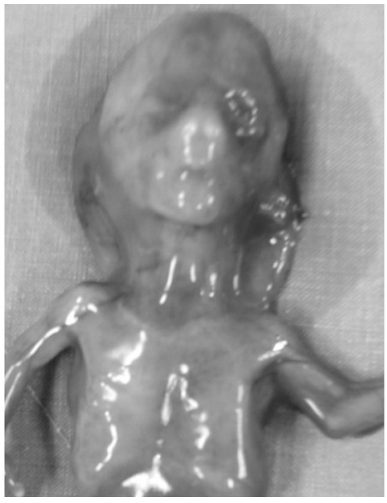

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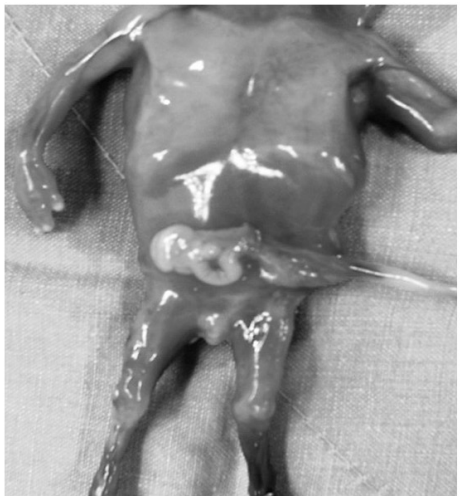

(D)

Fig. 1. Huge multiseptated cystic hygroma surrounding the fetal abdomen (A). Postnatal karyotype analysis reported 45,XO, Turner syndrome (B). Gross findings; Cystic hygroma surrounding the fetal cranium and neck (C), Gastroschisis : small and right-sided abdominal wall defect with protruded bowel (D).

Case report

A 24-year-old female, gravida 1, para 0, conceived spontaneously. At 14-weeks' gestation, a follow-up ultrasound demonstrated cystic mass of fetal neck. The patient was referred to our hospital at 15-weeks'gestation. On admission, ultrasound scan revealed thickened nuchal fold and huge multiseptated cystic hygroma surrounding the fetal cranium, neck and abdomen (Fig. 1). There was a finding of mild hydrothorax and ascites. It was suspected Turner syndrome or hydrops fetalis. After counseling, therapeutic abortion was performed by laminaria and misoprostol administration into the uterine cervix. Postnatal cytogenetic analysis for placental tissue revealed 45,XO (Fig. 1). Postnatal examination of the fetus revealed an additional gastroschisis with multiple bowel loops outside the abdomen, which was not detected by antenatal ultrasonography. It was confirmed a huge septated cystic hygroma surround the fetal neck on gross inspection (Fig. 1).

Discussion

Turner syndrome is among the most common cytogenetic errors when assessed shortly after conception, constituting 1–2% of all conceptions.[8] Turner syndrome affects about 50 per 100,000 female live births,[9] although only approximately 1% of fetuses with 45,X survive to term,[10] and as many as 15% of spontaneous miscarriages have a 45,X karyotype.[11] In approximately 50% of the cases, the affected individuals have a 45,X karyotype, whereas the others display various abnormalities of one of their sex chromosomes or may be mosaic.[12]

Although some studies suggest that Turner syndrome can be identified by a multiple biochemical marker screening,[13] the most useful tool in its noninvasive prenatal diagnosis is ultrasonography. Known fetal congenital anomalies in Turner syndrome cases were hygroma colli, hydrops fetalis, increased nuchal translucency (>3 mm), ventriculomegaly (>10 mm), cardiac defects (coarctation of aorta, ventricular septal defects, tetralogy of Fallot, dilated right ventricle), renal abnormalities, short femur (<10th percentile), choroid plexus cyst, echogenic bowel, echogenic intracardiac focus.[14] The most frequent anomaly associated with Turner syndrome was cystic hygroma. Possible mechanisms of this early-onset fluid collection of fetuses with Turner syndrome include abnormalities of the great arteries of the heart, abnormal development of the lymphatic system, and abnormal composition of the extracellular matrix of different tissues.[15,16]

In addition to cystic hygroma, some sonographic-features that are often seen in affected fetuses at the 11–14th weeks include tachycardia and early-onset fetal growth restriction.[17] In our case, estimated fetal age was 13-weeks' gestation on ultrasound scan at 15-weeks' gestation.

Gastroschisis is not associated with Turner syndrome or other aneuploidy. Gastroschisis is a congenital malformation characterized by an abdominal wall defect consisting in visceral herniation through a usually small wall defect lateral to an intact umbilical cord (almost always on the right side) and not covered by any membrane.[18] Other anomalies of the abdominal wall considered as completely different conditions from gastroschisis include omphalocele, limb-body wall complex (LBWC), and ectopia cordis. The reported total prevalence rate varies between countries from 0.5 up to 4 per 10,000 births.[18] In many countries the total birth prevalence rate is rising.[19] The association with young maternal age has been reported in many.[20,21] The explanation of these two consistent characteristics is still not fully understood.

The average gestational age at diagnosis for gastroschisis has been reported to be at mean 17 weeks of gestation, within a range of 15–19 weeks.[22] In our case, gastroschisis could not be detected by prenatal ultrasound, probably due to the large membrane of cystic hygroma surrounding the whole body, including the fetal abdomen. 15 weeks' gestational age might be early to diagnosis this malformation by transabdominal sonography. Transvaginal sonography was not performed because fetal anomalies associated with Turner syndrome was not found by transabdominal sonography.

There are possible explanations for the co-existence of gastroschisis with cystic hygroma might be compression of the cystic mass on the abdominal wall causing occlusion of the terminal segment of the right omphalomesenteric artery or the flow-related anomalies of the cardiovascular system commonly seen in Turner syndrome.[23]

Although gastroschisis tends to be an isolated anomaly not associated with Turner syndrome, there are rare cases of the literature of Turner syndrome co-existing with gastroschisis.

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