<table>
<thead>
<tr>
<th>Code</th>
<th>Title</th>
<th>Code</th>
<th>Title</th>
</tr>
</thead>
<tbody>
<tr>
<td>F1-01</td>
<td>Hypochondroplasia in a family with FGFR3 gene mutations: antenatal ultrasound findings.</td>
<td>F1-12</td>
<td>Paternal uniparental disomy of chromosome 16 resulting in hemoglobin Bart’s hydrops fetalis.</td>
</tr>
<tr>
<td>F1-02</td>
<td>Multiple pregnancy in a primigravida with uncorrected pentalogy of fallot.</td>
<td>F1-13</td>
<td>Analysis of the outcome of 200 cases of fetal isolated nasal abnormality at 11-13+6 weeks.</td>
</tr>
<tr>
<td>F1-03</td>
<td>Fetal intracranial teratoma: a case report. Pregnancy with thalassemia β /hemoglobin e disease.</td>
<td>F1-14</td>
<td>The outcome of cystic hygroma with aneuploidy and structural abnormalities vs euploidy in Women’s hospital in Qatar.</td>
</tr>
<tr>
<td>F1-05</td>
<td>Thanatophoric dysplasia type 1 with myelomeningocele: case report.</td>
<td>F1-16</td>
<td>Limb body wall complex: a case report.</td>
</tr>
<tr>
<td>F1-06</td>
<td>Arnold-Chiari type II malformation with myelomeningocele: a case report.</td>
<td>F1-17</td>
<td>The hemolytic disease of the fetus and newborn due to alloanti-M: three Chinese cases report and review of the literature.</td>
</tr>
<tr>
<td>F1-07</td>
<td>Difficulties in the antenatal diagnosis of ectopic kidney: A case report.</td>
<td>F1-18</td>
<td>Transillumination role as diagnostic tool of hydranencephally in limited facilities of rural hospital.</td>
</tr>
<tr>
<td>F1-10</td>
<td>A comprehensive management of gastroschisis at Sanglah General Hospital Bali.</td>
<td>F1-21</td>
<td>Holt-Oram syndrome: Analysis of Five Cases</td>
</tr>
<tr>
<td>F1-11</td>
<td>Acephalous fetus: a rare case.</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
Hypochondroplasia in a family with FGFR3 gene mutations: antenatal ultrasound findings.

WC Tan, DKL Chan, & HK Tan.
Department of Obstetrics and Gynaecology, Singapore General Hospital, Singapore.

CASE REPORT

- Patient was a 30 year old female, gravida 1 para 0. She had no family or medical history of note. She was of short stature, measuring 1.28m. She presented at our department at 29 weeks. Ultrasound scans were performed: (i) 29+2 weeks revealed short limbs; femur and humerus were below 3rd percentile. Right kidney contained multiple cysts. Female foetus. Amniotic fluid was normal, estimated foetal weight was 1028g, 5.3 centile. (ii) 31+4 weeks. Long bones were below 5th centile. Unable to demonstrate right kidney and right renal artery. A 1.5 cm cyst was demonstrated in right renal fossa. (iii) 34+1 weeks. Head circumference and abdominal circumference were within normal limits and Dopplers were normal. All long bones were below 5th centile with some frontal bossing, likely constitutional in view of maternal skeletal dysplasia. Right kidney was not visualised. A 2cm cyst was seen in right renal fossa - likely isolated right renal dysplasia. Postnatal examination of child revealed total length 44cm: upper segment 29cm, lower segment 15cm, arm span 40cm, flat nasal bridge, left clinodactaly, normal female genitalia, normal nose/mouth/ears. A diagnosis of hypo-chondroplasia, based on the FGFR3 test was done by pediatric geneticist.

- Two years later, patient had her second pregnancy. Ultrasound scan at 19 weeks noted all long bones at 5th percentile. No other fetal anomaly was detected, normal female genitalia. Postnatal examination revealed Upper segment: lower segment ratio 1.7, arm span 45cm, length <3rd centile. Clinically, posture was hypotonic with protubent abdomen; facial appearance looked similar to sister who was diagnosed with hypochondroplasia, frontal bossing; cranial ultrasound was normal. FGFR3 testing confirmed hypochondroplasia.

CONCLUSION: Although mutation in FGFR3 gene is a known cause of hypochondroplasia, researchers suspect that mutations in other genes are involved, although they have not been identified.
Multiple pregnancy in a primigravida with uncorrected pentalogy of fallot.

P Partana, JKH Tan, EL Tan, JL Tan, & LK Tan.
Department of Obstetrics and Gynecology, Singapore General Hospital, Singapore.

Pentalogy of Fallot is a cyanotic congenital heart disease that has guarded prognosis without surgical intervention in infancy. This condition refers to the co-existence of an atrial septal defect (ASD) and Tetralogy of Fallot, the latter of which comprises an over-riding aorta, obstruction of the right ventricular outflow tract (RVOT), right ventricular hypertrophy (RVH) and a ventricular septal defect (VSD). Despite this additional anatomical defect, the underlying pathophysiology of PoF and ToF remains similar. Women with uncorrected defects rarely survive into childbearing age and pregnancy in this group is associated with a high rate of perinatal loss. Physiological cardiovascular changes in pregnancy can lead to maternal haemodynamic instability with subsequent adverse cardiac sequelae with or without fetal decompensation. Optimum management and pregnancy outcomes in mother with uncorrected Pentalogy of Fallot and twin pregnancy have not been described in the literature. We describe a successful case of monochorionic diamniotic twin pregnancy in an affected woman who has not undergone surgical repair. Her pregnancy progressed without any adverse cardiopulmonary complications. Her caesarean delivery and postpartum recovery were favourable, with successful birth of two healthy babies at 35.7 weeks. This case emphasises the importance of a multidisciplinary team, especially of obstetricians with expertise in high-risk pregnancies, adult congenital heart disease cardiologists and anaesthesiologist.
Fetal intracranial teratoma: a case report.
DENSAK PONGROJPAW MD.
Maternal Fetal Medicine Unit, Department of Obstetrics and Gynecology, Faculty of Medicine, Thammasat University, Pathumthani, Thailand.

OBJECTIVE
The author report a case of fetal intracranial teratoma diagnosed at 26 weeks of gestation after prior normal sonographic examination.

MATERIALS & METHOD
Congenital intracranial tumors are rare and only account for 0.5-1.5% of all pediatric brain tumors. The diagnosis is usually made by ultrasonography in fetal life, as diagnosis of an intracranial mass, disrupting normal architecture with or without hydrocephalus. Teratoma is the most common tumor in neonatal period making up 30-50 percent of tumors.

RESULT
A 37-year-old woman, G1P0, 26 weeks of gestation was referred to Maternal Fetal Medicine Unit, Thammasat university hospital because of enlarged fetal head. Her antenatal record was uneventful. Ultrasonography revealed a 4.43 cm x 5.54 cms, heterogeneous intracranial fetal mass at the pituitary gland area. The enlargement of circle of Willis and macrocephaly were seen by colour Doppler and 3-D ultrasonography respectively (Figure 1-3). A postmortem histological examination showed an immature teratoma.

CONCLUSION
Massive congenital intracranial teratoma is an extremely rare neoplasm with a poor prognosis. They grow rapidly and cause extensive destruction in the brain.
BACKGROUND

Thalassemias are group of inherited autosomal recessive hematologic disorders that cause hemolytic anemia. Beta thalassemia is the result of deficient or absent synthesis of beta globin chains. Beta thalassemia trait (minor) occur if there is one gene defect on chromosome 11, which is usually asymptomatic. However, people who inherit combination of globin structure disorder such as hemoglobin E with thalassemia β or α, may have a serious hemoglobin disorder and cause severe anemia during pregnancy. Worldwide, patients with thalassemia beta/hemoglobin E (β-thal/HbE) represent 50% of severe beta thalassemia. The highest frequencies are observed in India and throughout Southeast Asia including Indonesia. In one study by Suchaya et al (2008), 0.2% of all pregnant women were affected by β-thal/HbE.

METHOD

Case Report

CASE

Patient 28 years old came first time at Obstetric Polyclinic Sanglah Hospital with diagnose first pregnancy 27-28 weeks + moderate anemia (Hb : 6.2 g/dL) + Thalassemia (β-thal/HbE).

During pregnancy, to improve fetal outcome she got three times blood transfusions. Fetal scanning already done at 34-35 weeks with result no major congenital was found in baby. Finally, at 37-38 weeks she delivered 2500 g male baby in vigorous condition. To determine types of thalassemia, already performed thalassemia DNA examination, which revealed her husband has normal DNA, her father carrier of HbE disease, whereas her mother carrier of minor β-thal. Therefore could be understand why she inherited β-thal/HbE disease.

DISCUSSION

Pregnancies affected by β-thal/HbE disease may be associated with a high rate of obstetric complications, especially fetal growth restriction, preterm labor and low birth weight. In this case, patient delivered healthy baby with no congenital anomaly, even though she suffered moderate anemia during pregnancy. DNA examination revealed that patient inherit β-thal/HbE disease from her parents.

KEYWORDS

Globin, Hemoglobin E, Thalassemia β.

REFERENCES

INTRODUCTION
Thanatophoric dysplasia (TD) is the most common lethal skeletal dysplasia occurred in neonatal.1 The typical clinical features are micrometric limbs, short costal bone, narrowed thorax, relative microcephaly, frontal bossing, midface hypoplasia, short vertebrae bones, abnormalities of the central nervous system which is the most lethal form of dwarfism in humans.1 Pulmonary hypoplasia resulting from fetal thoracic deformity can lead to severe respiratory insufficiency at birth, thus perinatal treatment becomes extremely difficult.2 As a result, early prenatal diagnosis is important because then early termination can be conducted so that minimize the risk to the mother. The improved ultrasound technology ease the observation of fetal development in the first trimester such as sex,3 facial bones4 and placental function.5

Thanatophoric dysplasia is very rare, ranging from 0.27 – 0.4 of 10,000 births, only a few are found in the first trimester in the case reported.6 Therefore, an easy early screening method for less experienced obstetricians remains a challenge.7

CASE REPORT
A 28 years old woman, pregnant with her second child, had the history of the first last menstruation in August 25th, 2017. During the first visit for routine prenatal examination using ultrasound in December 31st, 2017, no maternal complaint was present, no history of disease, normal delivery for the first child and the first child condition was normal and no abnormalities, no family history of congenital abnormalities was reported. In the ultrasound examination, the scoring of long bones with strong suspicion of TD was found (Figure 1). In the second visit, in January 24-25th, 2018 at an ultrasound examination found single pregnancy, life, biparietal diameter (BPD) of 4.86 cm consistent to the 20 weeks 5 days gestation, head circumference (HC) of 17.12 consistent to the 19 weeks 5 days gestation, while femur length (FL) of 1.18 cm consistent to the 13 weeks 3 days gestation. In the transverse section impression of the head, abnormal head image and frontal bossing (figure 2) were found. In the coronal section of the face, hypertelorism (figure 2a and 2b) was found. Impression of thorax was small (figure 3). Short impression of FL, short tibia and fibula and fingers were difficult to assess (figure 4). After counseling to both parents, pregnancy termination was conducted and male baby was obtained and found macrocephaly, a narrow bell-shaped thorax, protuberant abdomen, curved long bones, meningocoele on the cervical area and spine bifida was occur in the lumbar sacral region which was undetected during ultrasound examination. Genetic examination was not performed because the patient refused due to the limited cost.

DISCUSSION
Thanatophoric dysplasia, osteogenesis imperfecta and achondrogenesis are the 3 types of lethal skeletal dysplasia which are most commonly found. The occurrence of skeletal dysplasia is often associated with abnormalities in other organ systems.6 Thanatophoric dysplasia is one type of skeletal dysplasia, caused by the mutation of the fibroblast growth factor receptor-3 (FGFR3) gene, it is the lethal type of achondrogenesis. Genetic alteration is responsible for both types of TD which caused by excessive activation of the FGFR3 gene causing disturbance to bone growth.8 There are two types of Thanatophoric dysplasia, namely TD I and TD II.9 This case is Thanatophoric acrodysostosis type I because it shows a typical impression of short and crooked femur bones, tibia and fibula, so that the legs appear to be bent inward. This condition had been diagnosed at 12 weeks gestation by ultrasound examination but still monitored at subsequent examinations to further confirm the type of abnormality experienced.

In this case, the termination was done at 20 weeks 5 days gestation based on the consideration that this case is lethal abnormalities associated with ultrasound findings of fetal thoracic deformity that can lead to serious lung insufficiency. Most TDs are dead within hours of birth due to respiratory problems caused by poor chest cavity or due to brainstem compression.10

Ultrasound examination in second trimester pregnancy is important to evaluate the presence of congenital abnormalities. The fetal skeleton has been able to be visualized with two-dimensional ultrasound examination at 14 weeks gestation, and fetal femur and humerus measurements are considered as part of baseline examination for a second trimester ultrasound evaluation. Fetus showing a femoral or humerus length less than 5th percentile or > 2 SD from the mean of the second trimester (24 weeks) should be evaluated further by assessing all fetal skeletons and allow for genetic counseling.11 Ultrasound examination is particularly sensitive in establishing the diagnosis of prenatal skeletal dysplasia disorder. Careful observation is needed for each fetus to assess the overall bone starting from the head and ending in the feet.12 When long bone measurements are equal to or less than 5th percentile or > 3 SD below the mean, and especially if the head circumference is greater than 75th percentile, it requires further expertise due to suspected skeletal dysplasia. In addition, the leathery also needs to be assessed further by evaluating deeper the chest circumference to abdominal circumference or ratio of thigh length to abdominal circumference. The chest circumference ratio to abdominal circumference of less than 0.5 or the thigh length ratio to the abdominal circumference of less than 0.16 are strongly suspected as fetus suffering from skeletal disorder. Babies who are born and have skeletal dysplasia should be examined for clinical and radiological evaluation. Pathological examination of bone and cartilage especially of femur and humerus for histomorphological analysis is because many skeletal abnormalities are associated with significant recurrence risk.13

CONCLUSION
The general description of TD are very short femur bones, small bone, cleft palate, short fingers, small chest and polyhydramnios. The occurrence of body shortening is usually evident since week 12. Ultrasound screening is the safest and non-invasive method. The use of ultrasound in the first trimester resulting in the TD prenatal diagnosis accuracy can be detected earlier in pregnancy. The focus and emphasis should lead early malformation at 12 weeks gestation due to the fact that first-trimester detection for early diagnosis and timely intervention of congenital malformations optimizes the management of pregnancy, delivery time, and reduce the mental, physical, and psychological trauma of woman associated with late termination. Our ability to distinguish between leathery conditions is a key to parental counseling. A detailed postnatal examination of the neonate and a definitive confirmation of the underlying pathology are still essential for an accurate diagnosis and to determine the risk of recurrence in subsequent pregnancies. In addition, the results of the prediction can be used as subsequent pregnancy management.

REFERENCES
Arnold-Chiari type II malformation with myelomeningocele: a case report.

KP Bautista, & AR Teotico.
Department of Obstetrics and Gynecology, Manila Central University, Philippines.

• It is essential that every second-trimester fetal anatomic survey includes a complete evaluation of the spine. Spinal and cranial lesions may be identified and diagnosed early with the use of ultrasound screening examinations. Such is the case with the Arnold Chiari II malformation, a rare congenital deformity characterized by displacement of parts of the cerebellum, fourth ventricle, pons and medulla oblongata through the foramen magnum into the spinal canal.

• We report a case of a 33 year old multigravida, whose first baby was affected with anencephaly. In this present pregnancy, the fetus was prenatally diagnosed with the fetal congenital anomaly of Arnold Chiari II malformation, open spina bifida and myelomeningocele.

• The patient underwent fetal congenital anomaly scan, revealing a vertebral defect at the level of L3-S4, and a protruding mass over it, containing linear echogenic areas, surrounded by anechoic fluid, suggestive of myelomeningocele. Prenatal detection of myelomeningocele by fetal ultrasound was greatly improved by the detection of the Arnold Chiari malformation, which was more easily appreciated than directly visualizing the spinal defect. A thorough evaluation of the entire fetus was then done in order to determine associated conditions, which may influence outcome of pregnancy, route of delivery, and the need for postnatal surgery. The patient delivered term via normal spontaneous vaginal delivery to a live baby girl weighing 3000 grams. Meningocele repair and closure of rachischisis was performed on the second day of life, followed by a shunting procedure to address the hydrocephalus.

• This case shows that prenatal diagnosis of Arnold Chiari II malformation, spina bifida and myelomeningocele is of great importance since it allows time for comprehensive discussions, counseling and emotional support for the parents of affected infants. Early detection and evaluation enables timely anticipation of antepartum and intrapartum management and facilitates appropriate postnatal surgical care.
Difficulties in the antenatal diagnosis of ectopic kidney: A case report.

LSL Lee, TWC Tan, & DKL Chan.

Department of Obstetrics and Gynaecology, Singapore General Hospital Singapore.

Introduction: Prenatal diagnosis of abnormal or abnormally located kidneys may prove to be challenging. They may present as abdominal or even thoracic masses. The application of colour Doppler to trace its vascular supply and perfusion may not provide confirmatory results. We present a case of ectopic kidney which was diagnosed antenatally as isolated renal agenesis.

Case report: Patient was a 21 year old Vietnamese, gravida 1 para 0. She was referred from private clinic at 25+1 weeks gestation with history of CMV infection in Vietnam. Screening scan done showed a male fetus. Right kidney and right renal vessels were not seen on scan. A provisional diagnosis of absent right kidney was made. Estimated fetal weight at 27.4th centile.

Fetal anomaly scan done at 26+4 weeks gestation showed normal amniotic fluid index. Right kidney was not demonstrated and right renal vessels were not identified on colour Doppler. There was no structure resembling right kidney noted in the fetal abdomen, thus suggesting isolated right renal agenesis. Left kidney, left renal artery and bladder are seen well. There was no other fetal anomaly of note. Suggestion was made for referral to neonatology after delivery to exclude pelvic kidney.

Growth scan at 34 weeks gestation could not demonstrate right kidney on scan again.

Postnatal ultrasound showed the right kidney is small in size with a bipolar diameter of 3.3cm. It lies in a lower position than normal. The left kidney is normal in size with a bipolar diameter of 4.2cm. Renal cortical echogenicities of both kidneys are within normal limits. No focal lesion seen in the bladder.
An autopsy case of Potter syndrome: bilateral renal agenesis with club foot.
AR Lee, SM Kim, SE Park, JY Kim, YS Han, & IC Jung.
Department of Gynecology & Obstetrics Daejeon, St. Mary's Hospital, Catholic University, South Korea.

Introduction
Potter's syndrome is a rare condition occurring at a frequency of 1:2000 to 1:5000 fetuses. Males are affected more commonly. Here we present a case of Potter syndrome with bilateral renal agenesis with low-set abnormal ears, right ventricular hypertrophy with club foot diagnosed on autopsy.

Case report
A 28 year-old primipara woman was referred from local clinic at pregnancy 20weeks and 6days for decreased amniotic fluid index. She had no underlying disease or previous operation history except laparoscopic ovarian cystectomy done three years ago. She had never smoked cigarettes and exposed to drinking alcohols during pregnancy. Her BMI was less than 30 during pregnancy. Under transabdominal sonography, the amniotic fluid was nearly absent. On the day of arrival, amniocentesis and 300cc of amnioinfusion was done. The amniotic fluid normalized after amniotic infusion. Chromosomal study was performed by using Giemsa-Tripsin-Leishman Banding technique, it is confirmed to have no numerical or structural chromosome abnormalities under the microscope of 550 resolving power. The patient told her amniotic fluid was low since 18 weeks.
We performed target sonography. Estimated fetal body weight was approximately 323g at the top 20 percentile on the graph. Under doppler sonography, bladder filling and renal arteries were not visible. Lying down adrenal sign with empty renal fossa was seen and right ventricular hypertrophy was seen. The chest was 19weeks sized suggesting pulmonary hypoplasia. For religious reason, she decided to continue pregnancy. One week later, she admitted via emergency department due to labor pain with amniotic membrane rupture. A 350gram weighted stillborn male baby was delivered by spontaneous vaginal delivery.
Autopsy finding
There was no kidney, ureter, bladder. The stillborn baby showed low-set ears, suppressed mandible and club foot.
Amniotic fluid chromosomal analysis : normal 46,XY
Six months later, the patient got pregnant twin males naturally. And she had caesarean section due to preterm labor on 34+2weeks. The babies were 2.2kg, 2.3kg without anomaly.

Discussion
When the antenatal sonography shows severely decreased amniotic fluid with intact amnion, we have to suspect urinary anomaly. If severe oligohydramnios, non-visualization of the bladder and empty renal fossa is seen, it reflects bilateral renal agenesis. But poor sonographic resolution of severe oligohydramnios makes it difficult to diagnose the disease.
The Characteristics of Congenital Anomalies in a Tertiary Teaching Hospital: Sanglah Birth Defect Integrated Center (SIDIC) Program
Sanglah Hospital Bali, Indonesia
Giri Chandra, AAN Jayakusuma, AAG Putra Wiradnyana, 1 Wayan Megadhana, Made Bagus Dwi Aryana
Maternal Fetal Division, Departement of Obstetrics and Gynaecology Udayana University, Sanglah Hospital Bali, Indonesia

OBJECTIVE
Congenital anomalies are one of the causes of increasing numbers neonatal and infant morbidity and mortality. This study aim to measure the incidence and characteristics of pregnancy with congenital anomaly in Sanglah Birth Defect Integrated Center (SIDIC) program, Sanglah Hospital, Denpasar from January 1st 2017 to December 31st 2017. This research is expected to be a reference or as basis on conducting future advanced research on characteristics of the congenital anomalies at Sanglah Hospital Denpasar, Bali, Indonesia.

METHOD
This research was a descriptive study. Data was conducted secondary from medical records of 1 year period. All pregnancy with congenital anomalies recorded in SIDIC program, Sanglah Hospital Denpasar, Bali, Indonesia on 2017 are included.

RESULT
Number of expectant mother with congenital anomaly baby who delivered at Sanglah hospital was 57 in a period of 1 year 2017. We found 65 type of congenital anomalies in 1 year, most system affected in the newborns was gastrointestinal system 24.5% (16), central nervous system 23.0% (15), multisystem anomalies 20.0% (13), musculoskeletal system 6.1% (4), cardiovascular system 12.3% (8), miscellaneous 12.3% (8) and genitourinary system 1.5% (1). The majority of mothers 75.4% (43) were in maternial age 20-35 years, 21.1% (12) in maternial age >35 years and only 3.5% (2) in maternial age < 20 years. Related with gestational age, there were 12.3% (7) in gestational age 14 - 23+6 weeks, 15.8% (9) in gestational age 24 - 27+6 weeks, 36.8% (21) in gestational age 28-33+6 weeks and 35.1% (20) in gestational age >34 weeks. The mode of delivery : 52.6% (30) pregnancies was delivered by vaginal delivery while 22.8% (13) was delivered by sectio caesarea, the rest of the pregnancies 24.6% (14) was medical futility. Most origin mother with congenital anomaly baby referred from obstetrician in Bali (47.4%).

CONCLUSION
Handling infant with congenital anomalies should be start when expectant mother comes to have an antenatal care. Mother with high risk pregnancy should be fully screened and targeted diagnostic prenatal checks should be done to uphold the diagnosis of congenital abnormalities in pregnancy. Sanglah Hospital are developing SIDIC (Sanglah Birth Defect Integrated Center) program to be able to perform fetal intervention for infants with congenital anomalies from national to international levels.

Table 1: The Characteristics of Congenital Anomalies in Sanglah Hospital Bali

<table>
<thead>
<tr>
<th>Congenital Anomaly</th>
<th>Frequency</th>
<th>Percentage (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Anencephaly</td>
<td>5</td>
<td>7.7</td>
</tr>
<tr>
<td>Hydrancephaly</td>
<td>2</td>
<td>3.1</td>
</tr>
<tr>
<td>Microcephaly</td>
<td>1</td>
<td>1.5</td>
</tr>
<tr>
<td>Hydrocephalus</td>
<td>2</td>
<td>3.1</td>
</tr>
<tr>
<td>Dandy Walker malformation</td>
<td>4</td>
<td>6.2</td>
</tr>
<tr>
<td>Spina Bifida</td>
<td>1</td>
<td>1.5</td>
</tr>
<tr>
<td>Labiopatolatoschisis</td>
<td>3</td>
<td>4.6</td>
</tr>
<tr>
<td>Cutis dysmorphic</td>
<td>1</td>
<td>1.5</td>
</tr>
<tr>
<td>Esophageal Atresia</td>
<td>4</td>
<td>6.2</td>
</tr>
<tr>
<td>Duodenal Atresia</td>
<td>4</td>
<td>6.2</td>
</tr>
<tr>
<td>Diaphragmatic Hernia</td>
<td>1</td>
<td>10.5</td>
</tr>
<tr>
<td>Omphalocele</td>
<td>1</td>
<td>1.5</td>
</tr>
<tr>
<td>Gastrochizis</td>
<td>5</td>
<td>7.7</td>
</tr>
<tr>
<td>Fetal Cardiac anomaly</td>
<td>8</td>
<td>12.3</td>
</tr>
<tr>
<td>Polycystic kidney disease</td>
<td>1</td>
<td>1.5</td>
</tr>
<tr>
<td>Fetal Hydrops</td>
<td>8</td>
<td>12.3</td>
</tr>
<tr>
<td>Multiple congenital anomaly</td>
<td>13</td>
<td>20.0</td>
</tr>
<tr>
<td>Total</td>
<td>65</td>
<td>100</td>
</tr>
</tbody>
</table>

REFERENCE
INTRODUCTION

- Gastroschisis from the Greek, means “belly cleft.” The features of this rare condition are a defect in the fetus abdominal wall which is extra-umbilical in location with no membranous covering the eviscerated mass of intestines and a normal umbilical cord insertion into the abdominal wall that is not involved with the evisceration.
- Gastroschisis is a full-thickness defect in the abdominal wall usually to the right of a normal insertion of the umbilical cord into the body wall. Gastroschisis occurs in 1 in 4,000 live births.
- Ultrasonographic visualization finding usually there is “freely floating loops” of bowel within the amniotic fluid with an abdominal wall defect to the right of the insertion of the umbilical cord at any point after the normal embryonic return of the intestine to the abdominal cavity at 10 weeks of gestation.
- Complex gastroschisis associated with bowel complications including intestinal atresia, perforation, necrosis or volvulus.
- Prenatal ultrasound markers like small for gestational age, intra abdominal and extra abdominal bowel dilatation (>6 mm), thickened intestinal wall and stool dilatation were correlated with poor outcome.

CASE

A 24 years old, Maternal Fetal Medicine Outpatient Clinic Sanglah General Hospital Bali at 26 weeks gestational age, Known gastroschisis since 18-19 weeks of gestational age at C&G and has been performed amniocentesis with karyotyping result was 46 XX, no gene abnormality.

Medical history: There are no history of gestational diabetes mellitus, gestational hypertension, vaginal discharge and vaginal bleeding. The father was smoker-smoking 10 cigarettes per day.

Obstetrics examination were within normal limit, fundal height appropriate with gestational age. From antenatal ultrasound we found full-thickness defect in the anterior abdominal wall located to the right of the intact umbilical cord. There was a frequency of bsupporting through the defect, floating in the amniotic fluid, which may be disproportionately large relative to the small size of the abdominal cavity.

DISCUSSION

- Gastroschisis remains as one of the congenital abdominal wall defect.
- There were several theories contain possible pathologies such as a vascular insufficiency associated with right umbilical vein or right omphalomesenteric artery, a rupture occurring on the bottom of umbilical cord during the physiologic herniation of intestines.
- The primer cause for the appearance of the anomaly is the inconsistency of the interaction between endoderm and mesoderm which is normally supposed to compose anterior abdominal wall in the end of the 3rd week of intratissue development.
- However, it is indicated that being a mother in a very young age rises and smoker or passive smoker increase risk of gastroschisis.
- It is shown that smoking increases the risk of encountering abdominal wall defect by 2.1 times.
- The defect on anterior abdominal wall is a 2-4 cm sized full-thickness abdominal wall defect located on mostly right, rarely left lateral side of umbilical cord.
- Frequently intestines and sometimes stomach, colon herniate towards the outside of abdomen.
- The organs herniating outside are not covered by membrane. Due to this fact and chemical effect of amniotic fluid, the intestines are inflamed.
- Fetal ultrasonography brings significantly benefit to diagnosis. Valid USG results could only be achieved after 14th-17th gestational week. Ultrasound image of gastroschisis is seen as small anterior abdominal wall defect located right lateral side of the umbilical cord and intestines draining from this area to amniotic space.
- There is no consensus about delivering babies antenatal diagnosed with gastroschisis by planned C-section. However, delivering the baby by C-section cause reduction of the risk of intra-abdominal organ trauma and infection.
- If there are no problems associated with fetal distress or intestinal damage, delivering the baby in around 39th week could be relevant.
- Treatment modalities such as primary closure, developing a ventral hernia to be closed soon after C-section are preferred recently. The prognosis is considered as good because of the frequency of having associated anomaly is low among the babies with gastroschisis. The mortality rate is known as between 5%-15%, mean 7.7%, provided respiratory circulatory insufficiency, sepsis or complications regarding total parenteral nutrition were not encountered. The survival rate is stated as 96% in cases with isolated gastroschisis treated in sufficient centers.

CONCLUSION

Improved understanding of gastroschisis, its early diagnosis by prenatal ultrasound, safe delivery of the fetus with a ventral wall defect, advanced surgical techniques for the reduction and intensive care management of neonates reduces the morbidity and mortality.

REFERENCES

Acephalous Fetus: A Rare Case Report

Akhmad Khalief, Rizki Azenda, Sandhy Prayudhana, Eddy Toynbee, Wiku Andonotopo

Department of Obstetrics and Gynecology Faculty of Medicine University of Indonesia, Department of Obstetrics and Gynecology Tangerang General Hospital

Introduction

Fetal acephalous is an extremely rare case which is defined by complete absence of the head. There has been no report regarding such case before.

Objective

The aim is to report a novel case and discuss about the process causing the fetal defect, and the mechanism of fetal behavior.

Case Report

A 27-year-old patient, gravida 2, parity 1, was referred to our center at 33 weeks of gestation. There was no relevant medical history and she was taking no medication, no history of smoking, drugs, or alcohol. She had no history of low intake, chemical hazards and pollution exposure. She had a low standard of prenatal care, as she had no first trimester ultrasound or other proper prenatal diagnostics in early pregnancy. She was then evaluated using 3D and 4D ultrasound, which confirmed the diagnosis of an acephalous fetus. (Figure 1 and Figure 2). CTG showed abnormal findings with low variability and no acceleration.

The fetal organs were normal. Both arms were normal but remained in the same position (in front of the chest). Bilateral clubfeet was found with more complex and frequent leg movements. A 3D CT-scan was performed (Figure 3). Due to patient belief, she wants to postpone the delivery until term pregnancy. Patient was admitted to emergency department at 34-35 weeks of gestational age due to premature rupture of membrane seven hours prior to admission. An emergency C-section was performed and the baby was delivered by shoulder extraction.

Baby girl was born, 1000 grams, 42 cm with APGAR score 5/3. The baby had a pointed cervical vertebrae with an opened end (Figure 4). Both arms were in front of the chest and only had few movements. Both legs showed more rigorous and frequent movement. Clubfeet were noticed. Bradycardia was noted soon after delivery and the baby died 2 hours later.

Discussion

Birth defects can be caused by single gene defects, chromosomal disorders, multifactorial inheritance, environmental teratogens, and micronutrient deficiencies.

This case came without proper examination in the early and mid-trimester of pregnancy, which has made tracing of the etiology very difficult. There is no literature showing chromosomal anomaly or even a single gene disorder as its origin. This new case could be classified as new gene mutation because no case has been reported in any journal before.

The structural development of the fetal head occurs between the 3rd and 8th weeks of gestation. In this case, impairment of neural crest formation at anterior pore might be caused by impairment or gene mutation in induction of cranial neural crest formation process. Consequently, the migration of neural crest to specific cells as formation process of head, face, and also scalp didn’t occur.

How the baby could survive for 2 hours and move for 15 minutes after delivery was an interesting question. Almost all the body’s mechanisms is controlled by the CNS in which the spinal cord performs the rhythmic and sequential activation of muscles in locomotion and resulted from complex and dynamic interactions.

The Central pattern generator (CPG) within the lumbosacral spinal cord segments provides the basic locomotor rhythm and synergies by integrating commands from various sources. This spinal circuitry can function independently in the absence of input from the brain to generate stable posture and locomotion and even activity to match changing conditions (e.g. stepping over obstacles). In the case of an acephalous fetus, the spinal circuit below the brain site seemed to stay active instead of became silent and worked in a modified manner.

Conclusion

In this very rare case, the baby could live and move without the head for two hours. The Movement can be explained by independent functioning of the CPG in the lumbosacral spinal cord.

References

α-Thalassemia is inherited as an autosomal recessive disorder characterised by a microcytic hypochromic anaemia. It is one of the most prevalent genetic diseases in the world and is especially frequent in tropical regions, including South China. An estimated carrier rate of 3.16%–11.72% for a-thalassemia has been reported in Guangdong Province of China. Universal prenatal screening for thalassemia by mean corpuscular volume (MCV) and hemoglobin electrophoresis is an integral part of prenatal care in Guangdong Province. Here we report an unusual case of Hb Bart’s disease.

◆ A 36-year-old, G4P1, Chinese woman had a low MCV and HbA₂(2.37%) in prenatal screening. Multiplex gap PCR for α-thalassemia – SEA, –α 3.7, –α 4.2, –α THAI and –α FIL deletions showed that the woman had normal α-globin genotype and her husband was heterozygous for α-thalassaemia-1 (– SEA ) deletion. The first trimester NT was 2.2mm. NIPT results for trisomy 21, 18, 13 were low risk. Ultrasound examination at 27+4 weeks’ gestation showed fetal cardiomegaly (cardiothoracic ratio was 0.48), pericardial effusion and raised middle cerebral artery peak systolic velocity (MCA-PSV) of 67.3 cm/s (1.9 MoM). Cordocentesis showed fetal anaemia with an Hb of 7.8 g/dl and Hb Bart’s disease. Multiplex gap PCR showed that the fetal was homozygous for α-thalassaemia-1 (– SEA ) deletion.

◆ Investigations into the cause of Hb Bart’s disease with multiplex fluorescent PCR using 10 STR markers spanning the whole Chromosome 16 showed paternal uniparental disomy.

◆ Our case suggests that Hb Bart’s hydrops fetalis with homozygous mutation in a fetus of a couple in whom only one parent is heterozygote of the alpha1 globin gene deletion should be further evaluated for possible UPD(16), initially by looking for cardiothoracic ratio and MCA-PSV through ultrasonography. With fetal Cardiomegaly or fetal anemia, obstetricians should remain alert for fetal Hb Bart’s disease.
Objective: To evaluate the true positive status of fetal isolated nasal abnormality at 11-13+6 weeks by ultrasound.

Methods: A retrospective analysis of the follow-up results of 200 cases of fetal solitary nasal bone abnormalities at 11-13+6 weeks was conducted. The development of the nasal bone in the middle pregnancy was compared with that in the early pregnancy. The 200 fetuses were divided into two groups. The true positive rate of nasal bone dysplasia between the two groups, the fetal crown-rump length <56.2mm group (Group A) and the fetal crown-rump length >=56.2mm group (Group B), was compared.

Results: 178 out of the 200 cases were normal nasal bone cases (89%). 22 cases of nasal bone dysplasia were confirmed in 200 patients with 20-24 weeks ultrasonic examination; the true positive rate was 11% (22/200). There were 5 cases of nasal bone dysplasia in the group A; whereas there were 17 cases of nasal bone dysplasia in the group B. The true positive rate for the Group A and the Group B was 4.39% (5/114) and 19.77% (17/69) respectively. The true positive rate of the two groups was statistically significant (P=0.0006).

Conclusion: There is still a big chance that the fetal nasal bone will develop completely in 20 weeks even if the 11-13+6 week ultrasound diagnosis shows the isolated nasal bone is abnormal. However, the positive rate of abnormal nasal bone is increasing as the increase of the examination of the gestational weeks.
The outcome of cystic hygroma with aneuploidy and structural abnormalities vs euploidy in Women's hospital in Qatar.

A Al-Ibrahim, & AMH -Baloushi.
Department of Obstetrics and Gynecology, Women's hospital-Hamad Medical Corporation, Qatar.

Introduction: Cystic hygroma (CH) incidence is approximately 1/6000 live births. 70–80% occur in the neck, remainder 20–30% occurs in the axilla, superior mediastinum, chest wall, mesentery, retro-peritoneal region, pelvis and lower limbs. CH is abnormal fluid accumulation in fetal neck, sometimes associated with aneuploidy, ultrasound screening increased CH detection.

As noticed in some published articles early cessation of gestation might affect intrauterine anomalies detection and possible outcome in viable fetuses, due to possible religious or cultural reasons we have good exposure to continuation in viable pregnancy with CH in consanguineous marriage.

Objective: To study perinatal screening of fetal CH, the associated aneuploidy and outcome in consanguineous marriage.

Material and Methods: Retrospective study conducted from reviewing computerized medical records of fetal CH, diagnosed by Fetomaternal medicine physicians in Hamad medical corporation, Qatar, between Jan 2015 and Jun 2017. Fetal sonographic examination and Karyotyping were performed. pregnancy and pediatric outcome data were collected from medical records. Analyzed by Wizard software with p value 0,05 as cut of for significance.

Results: 114 cases were included. Mean maternal age was 31.6±1.4, 39.5% were nationals. Consanguinity present in 50%. CH discovered at mean gestational age of 12.6±0.6 weeks, 60.5% were septated., 68.4% had prenatal karyotype; of them 48.6% were normal. 12 cases traveled abroad, of the remaining 102 patients, 64 patients were missed miscarriage at mean gestational age of 15.8±0.6 weeks, 2 spontaneous miscarriage and 9 cases were IUFD at median gestational age of 26.9. Remaining 27 cases continued the pregnancy, 25 delivered, and 2 cases terminated for scar dehiscence and sever structural abnormalities.

Conclusion: CH has poor outcome, but couples can be encouraged to complete pregnancy especially if fetus is euploid and structurally normal with future risks of neuro-developmental delay, this continuation is missed in other studies and available in ours.
Intrauterine Transfusion for fetal Anemia: Indications and Implications

Mandakini Pradhan, Neeta Singh, Sangeeta Yadav
Department of Maternal and Reproductive Health, SGPGIMS, Lucknow, Uttar Pradesh, India 226014
mandakini_pradhan@rediffmail.com

INTRODUCTION

- Transfusion of red cells into fetus: one of the most successful fetal therapeutic procedures
- Most common cause of fetal anemia is Red cell isoimmunisation; most common being sensitization against RhD antigen
- Other rare causes of fetal anemia requiring fetal transfusion include Parvovirus B19 infection, placental chorangioma, twin to twin transfusion
- Fetal transfusion can be intravascular at cord insertion, free loop or intrahepatic umbilical vein, intra peritoneal and intracardiac site
- Fresh O negative, irradiated, leukocyte free PRBC with 75-85% hematocrit is used

MATERIALS AND METHODS

- Retrospective analysis of Intrauterine transfusion between January 2010 to December 2011
- A total of 362 fetal transfusion in 126 patients was done. In 124 patients fetal transfusion was done for Red cell isoimmunisation and in 2 cases fetal transfusion was done for non immune causes, 1 each for parvovirus B19 infection and 1 for placental chorangioma.
- Site of transfusion was intravascular in all except one which was intraperitoneal.
- Requirement of multiple transfusions, fetal presentation (hydropic vs non-hydropic), overall neonatal survival, maternal and fetal complications were analysed.

RESULTS

- Intravascular placental insertion route was used in 352 transfusions. Anterior placenta was present in 61.9% cases and 38.8% had posterior placenta. Rest nine it was done in intraperitoneal portion of umbilical vein.
- Average number of transfusion required for each patient was 1.9 (range 1-7). Mean period of gestation at time of first transfusion was 28.3 weeks, mean gestational age at delivery was 34.2 weeks.
- Out of 124 patients requiring transfusion due to immune cause, Anti D was present in 92 cases, Anti D and Anti C was present in 25 cases and 7 case were due to multiple antibodies along with Anti D.
- Needle dislodgement was noted in 34 procedures, leak from puncture site was noted in 9 procedures, fetal bradycardia was seen in 7 procedures, 1 each of abortion, cord spasm.
- Least hematocrit at which transfusion was given was 3.9% and earliest gestational age at which intravascular transfusion was done was at 18 weeks

Demographic data

<table>
<thead>
<tr>
<th>Parameter</th>
<th>Value</th>
</tr>
</thead>
<tbody>
<tr>
<td>Mean Age</td>
<td>29.1 years (22-40 years)</td>
</tr>
<tr>
<td>Mean gestation at which first intravascular transfusion given</td>
<td>28.3 weeks (18-36.6 weeks)</td>
</tr>
<tr>
<td>Mean gravidity</td>
<td>4.8 (2-9)</td>
</tr>
<tr>
<td>Mean gestational age of delivery</td>
<td>34.2 weeks (22-39.3 weeks)</td>
</tr>
<tr>
<td>Mean number of transfusion</td>
<td>2.9 (1-7)</td>
</tr>
</tbody>
</table>

CONCLUSIONS

- Intrauterine transfusion is safe and effective procedure to treat fetal anemia for different indications.
- The most common indication is Red cell Rh D isoimmunisation
Limb body wall complex: a case report

Gatita Iswaranti*, Rizki Azenda**, Wiku Andonotopo***
*Department of Obstetric and Gynecology/Faculty of Medicine-Universitas Indonesia, Ciptomangunkusumo Hospital
**Department of Obstetric and Gynecology, Tangerang General Hospital

Introduction
Limb body wall complex (LBWC) is a rare complicated sporadic polymorphic fetal malformation syndrome, characterized by a wide spectrum of severe anomalies in the body wall. The incidence at birth is about 0.32 per 100,000 births because the majority of affected fetuses undergo intrauterine deaths.1,2,3

Case Report
A 22-year-old, G1P0A0 presented to the obstetrics outpatient department in Tangerang General Hospital in the second trimester of her first pregnancy for a routine antenatal check up. History of hypertension, diabetes, asthma and allergy were negative. There was no history of any drug intake except for iron and folic acid supplementation. Her routine hematochemical and biochemical investigations were within normal limits.

A routine anomalous scan done at 18th trimester showed a fetus with features of the limb body wall complex syndrome. The cranium and fetal brain ultrasound showed anencephaly. While the ultrasound of the fetal thorax and abdominal region showed ectopic cardiac and liver and gastrochisis. Scoliosis were also found. Both upper and lower extremities were visualized and found club foot. There were no oligohydramniius found.

After c-section, we found the fetus weighed 1520 gram. On external examination a number of anomalies were noted. The genitalia were ambiguous. Encephalocele was seen over the head of the fetus in the occipital region. The most striking abnormality was a left sided anterior abdominal wall defect, from which gastrointestinal organs were protruding. The organs identified included the liver and the intestine. A defect in the posterior abdominal wall was also noted. A number of skeletal abnormalities were noticed. There were scoliosis and amelia of the digits of the right upper limb.

Discussion
The limb body wall complex is also known as the body stalk syndrome. It is a rare entity characterized by severe malformations. Most fetuses are aborted, either spontaneously or by medical means. Most of the remaining are stillborn. Postnatal survival for a significant duration is extremely rare. Traditionally diagnosis has been based on the Van Allen et al. criteria, i.e. the presence of two out of three of the following anomalies:
1. Encephalocele or encephalocele with facial clefts
2. Thoraco and/or abdominoschisis
3. Limb defects

Conclusion
Limb-body wall complex is a lethal polymorphic fetal malformation syndrome. The exact underlying cause of limb-body wall complex (LBWC) is currently unknown. There is no specific treatment for this condition. Postnatal survival for a significant duration is extremely rare. It is important to educate parents about the child’s prognosis and outcome. Prenatal diagnostic screening for fetoscopy and anomalies should be done at midtrimester.

References
4. AAP 87.
### Hemolytic disease of the fetus and newborn due to alloanti-M: three Chinese case reports and a review of the literature

Si Li, Chunyan Mo, Linhuan Huang, Xiaomei Shi, Guangping Luo, Yanli Ji, Qun Fang

### Objective
An increasing number of cases suffering from severe hemolytic disease of the fetus and newborn (HDFN) caused by alloanti-M, has been reported mainly in the Asian population. The objective of this abstract is to describe the clinical characteristics of alloanti-M-induced hemolytic disease of the fetus, along with a review of relevant studies or cases published in English and Chinese.

### Methods
Three pregnant Chinese women with a history of abnormal pregnancy with hydrops fetalis were encountered. During this pregnancy, a series of clinical examinations and an alloantibody identification against red blood cells (RBCs) and platelets were conducted. Intrauterine transfusion (IUT) and postnatal transfusion were then performed in the fetuses. In addition, the HDFN cases caused by alloanti-M reported in different ethnic groups and the clinical and serologic features are also summarized.

### Results
Three pregnant women were identified with an NN phenotype and IgM mixed with IgG alloanti-M in serum. Their fetuses were found severe anemia by cord blood testing. Additionally, IgG alloanti-M were detected in the cord blood of the three fetuses with titers ranging from 1:1 to 1:128. Moreover, low reticulocyte counts and negative direct antglobulin test were also shown in two of the fetuses. After receiving IUTs and postnatal transfusion, these three fetuses eventually survived and healthfully developed. A total of 67 fetuses/neonates with HDFN caused by alloanti-M were reviewed from 21 articles in English and 40 articles in Chinese.

### Conclusions
MN alloimmunization was the most common cause, next to Rh alloimmunization, resulting in severe HDFN, which is often seen in hydrops fetalis in the Asian population.

### Table 1. The Clinical findings of three pregnant Chinese women and affected fetuses due to alloanti-M

<table>
<thead>
<tr>
<th>Case</th>
<th>1st GA</th>
<th>Times</th>
<th>Hydrops</th>
<th>Hbmin (g/L)</th>
<th>Hct</th>
<th>Ret (10^9/μl)</th>
<th>Ret %</th>
<th>RPI %</th>
<th>GA</th>
<th>Sex</th>
<th>Tx</th>
<th>Major Complications</th>
<th>Outcomes</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>20&lt;sup&gt;4&lt;/sup&gt;</td>
<td>8</td>
<td>No</td>
<td>23</td>
<td>0.077</td>
<td>138.1</td>
<td>16.8 %</td>
<td>1.1 %</td>
<td>35</td>
<td>M</td>
<td>3</td>
<td>ABO incompatibility</td>
<td>Alive</td>
</tr>
<tr>
<td>2</td>
<td>31&lt;sup&gt;1&lt;/sup&gt;</td>
<td>2</td>
<td>No</td>
<td>77</td>
<td>0.217</td>
<td>84.2</td>
<td>4.1 %</td>
<td>1.1 %</td>
<td>35</td>
<td>M</td>
<td>2</td>
<td>NEC, icterus</td>
<td>Alive</td>
</tr>
<tr>
<td>3</td>
<td>27</td>
<td>5</td>
<td>Yes</td>
<td>23</td>
<td>0.009</td>
<td>/</td>
<td>/</td>
<td>/</td>
<td>36</td>
<td>F</td>
<td>3</td>
<td>/</td>
<td>Alive</td>
</tr>
</tbody>
</table>

1st GA: the 1st gestational age of intrauterine transfusion(IUT); Hbmin: the lowest hemoglobin before IUT; Ret: reticulocyte count before IUT; Ret%: reticulocyte percentage before IUT; RPI: Reticulocyte Production Index, GA: gestation age at birth; Tx: times of transfusion after birth; NEC: necrotizing enterocolitis.

Corresponding to: Si Li
Fetal Medicine Center, Department of Obstetrics and Gynecology, The First Affiliated Hospital of Sun Yat-Sen University, Guangzhou, China, 510080
Email: grace980050@gmail.com
Transillumination: Diagnostic Tool of Hydranencephaly In Limited Facilities
Ilham Utama Surya, Sri Pudyastuti*, Azen Salim**, Shindy Octaviana, Gunung Mansell
Malingping Hospital, Persahabatan Hospital*, Obstetric and Gynecology Department Cipto Mangunkusumo Hospital**

INTRODUCTION
Hydranencephaly was the absent of cerebral cortex completely with skull intact. It was replaced by cerebrospinal fluid, glial tissue, and ependyma. However, it still had midbrain, cerebellum, thalamus and choroid plexus. This rare condition had incidence between 1 in 4000 and 1 in 10,000 births. Hydranencephaly was considered the most severe form of a spectrum of disorders that includes porencephalic cyst and schizencephaly. The pathophysiology of hydranencephaly was obstruction of the internal carotid artery that leads to brain damage. The risk factors include infection, maternal condition and genetic disorder. Infection such as toxoplasmosis, cytomegalovirus, herpes simplex was associated to it. Hemorrhagic states such as familial factor XIII deficiency had also been reported. A variety syndromes had been reported in association with hydranencephaly including trisomy 13, agenesis malformation complex, lethal multiple pterygium syndrome, and both renal aplasia dysplasia and polyvacular development heart defect. Hydranencephaly may occur in a surviving co-twin following single intrauterine fetal demise in a monochorionic twin. It had also been reported in association with a primary congenital rhadoleic tumor of the brain.1

Deficit neurology such as seizures, vision difficulties, exaggerated muscle reflex, increased muscle tone, delayed development, poor body temperature, breathing and digestive problem occurred in infant period. Almost half of the infant with hydranencephaly die within a month and fewer than 15% survive 1 year. In some case series patient were alive until age 9, 12, 17 and 29 years. The diagnosis was confirmed post-natal via MRI, CT Scan and ultrason. CT scan was helpful in distinguishing hydranencephaly from extreme hydrocephalus and might be demonstrate maximal preservation of the frontal cortex, and a normal cerebral vascular architecture in hydranencephaly. MRI was the most definitive imaging technique for the diagnosis of hydranencephaly. However, in limited facilities those modalities could not be used. Therefore, alternative modality was necessary.1

MATERIAL AND METHODS
Case illustration and literature review of the case that was found during emergency setting in Malingping Hospital

CASE ILLUSTRATION
A 19 years old women G1 term pregnancy came to the emergency room due to prolonged stage of labor. The patient conducted ANC 4 times to the midwives and never had been ultrasound before. IVP revealed term pregnancy, she had simultaneously contraction, there was no water broke and hypertension. During pregnancy she had no complain, took no drugs, and no genetic disorder in the family. Physical examination revealed vital sign were normal, general status were normal, obstetric status fundal height 38 cm, the below presentation was the head, and identified hand palpable 3 fingers above the symphysis, contraction 3x/10 minutes/45 second. Vaginal touch revealed head in the hodge 2, amnion membrane intact, both femur were palpable, full dilatation. Ultrason show anechoic structure with intact skull with the present of cerebellum and brainstem. The diagnosis was dystocia second stage of labor with fetal hydranencephaly. Emergency C-section was performed, born baby boy 3800 g, 54 cm, AS 9/10. The day after, We decided screening... The baby was entered in the dark room and given light with high intensity, resulting fluorescent.

DISCUSSION
Transillumination of the skull was the traditional method of postnatal diagnosis of hydranencephaly. However, transillumination may also been with extreme hydrocephalus. Transillumination of the skull was first described in 1831 by Richard Bright and was later recognized as the first form of light-based diagnosis of hydrocephalus. Over time the technique was modified and used to diagnose intracranial hemorrhage in the neonate before the availability of ultrasonography. It had since been used as a screening procedure for infants with macrocephaly and those suspected of having subdural hematoma, hydrocephalus, hydranencephaly, porencephaly, increased intracranial pressure and even skull fracture and nutritional deficiencies.2

Attempts have been made to develop standardized light source and measurement standards based on age. In general, when using a standard 2 cell flash light held light to the anterior fontanel, transillumination of more than 2 cm around edge of the beam at 30° of the transillumination suggest underlying pathology. However, depending premature, age, light source and operator technique. Given it low cost and simplicity to apply, transillumination was a useful initial screening test for infants with abnormal findings in limited facilities.2

CONCLUSIONS
Transillumination could be performed when there are no imaging modalities such MRI and CT Scan. However, it could not differentiate hydrocephalus.

REFERENCES
Prenatal diagnosis of four cases of Cri-du-chat syndrome.

ASL Mak, TWL Ma, WH Lau, & KY Leung.
Department of Obstetrics and Gynaecology, Queen Elizabeth Hospital, Hong Kong.

Introduction: Cri-du-Chat syndrome is rare, caused by variable size deletions in the short arm of chromosome 5, and characterized by many significant phenotypes and psychomotor retardation. Prenatal diagnosis of this syndrome by ultrasound can be difficult. We aim to report cases of Cri-du-chat syndrome detected by other means including combined first trimester screening (cFTS) and cell-free DNA (cfDNA) testing.

Methods: Database of our prenatal diagnostic clinic from July 2010 to February 2018 was searched for cases of prenatally diagnosed Cri-du-chat syndrome. cFTS and cfDNA testing results, ultrasound features, invasive prenatal test results, outcomes of pregnancy were traced and analysed.

Results: From a total of 9,816 patients consisting mainly of Chinese, we found four cases of prenatally diagnosed Cri-du-chat syndrome. Prevalence was around 1 in 2,544 among high-risk group and around 1 in 12,500 on the whole. Three patients had termination of pregnancy.

Two cases had prenatal ultrasound abnormalities including small cerebellum, prominent lateral ventricles or cysterna magna. In one of them, prominent renal pelvis was also found in the first-trimester scan. There were no abnormal ultrasound findings in the other two cases, although examination after termination of pregnancy showed subtle features: low-set ears in one and a triangular face in the other.

In one case, the first sign was positive cFTS with high risk of trisomy 18 and low pregnancy associated plasma protein-A (PAPPA).

cfDNA testing was positive in one patient but negative in another. cfDNA testing in the former showed reduced amount of DNA in 5p; whereas cfDNA testing in the latter showed a low risk in 5p deletion. Subsequent karyotyping of the former showed a 10.50Mb copy loss in 5p15.33-p15.2; while the latter had a pathogenic 10.14Mb copy loss in 5p15.33-p15.2.

Conclusions: Cri-du-chat syndrome can be presented prenatally as abnormal findings of ultrasound, cFTS or cfDNA.
The value of ultrasonography in prenatal diagnosis of criss-cross heart.

XY Lin.
Department of Ultrasound, Bao'an Matenity & Child Health Hospital, Jinan University, China.

Objective: To explore value of ultrasonography in diagnosis of prenatal criss-cross heart (CCH).

Method: We summarize the characteristics of echocardiography and the key points for differential diagnosis of CCH by reviewing 2 cases identified in the Shenzhen Bao'an Maternity& Child Health Hospital. Ultrasonographic investigation were as following: (1) The typical four-chamber view of the heart was not able to be revealed in transverse plane of the fetal chest. (2) A four-chamber view seen in the sagittal plane of the fetal chest in which left and right ventricles were arranged up and down while the ventricular septum was horizontal. (3) Scanning from the upper abdomen to the chest cavity showed that left and right ventricle inflow channels were arranged in a criss-cross pattern. Most of the left ventricular inflow was from the left rear to the right front and a few from the right rear to the left front. Most of the inflow of the right ventricle was from the right rear to the left front, while a few was from the left rear to the right front. (4) In the transverse plane of the fetal chest color Doppler ultrasound displayed the criss-cross arrangement of the inflow tracts into the two ventricles.

Results: Case 1 is an infanta fetus of 23 weeks and fetal heart sound image revealed dextrocardia, CCH, single arterial trunk, severe pulmonary stenosis and ventricular septal defect; Case 2 is an infant fetus of 23 weeks and heart sound image demonstrated dextrocardia, CCH, aortic arch stenosis and ventricular septal defect. Both cases were confirmed by pathological anatomy after labor inductiontermination of pregnancy. Fetal myocardial tissue of one case had a gene chip test and itsrevealed normal result was normal by chromosomal microarray analysis.

Conclusion: Prenatal ultrasound is valuable to the diagnosis fetal of fetal CCH as it can display in the sagittal section of the thorax the four-chamber view which cannot be viewed clearly in the transverse plane of fetal chest.
Holt-Oram syndrome: Analysis of Five Cases

Abstract:
Background:
Holt-Oram syndrome (HOS) is an autosomal dominant disease caused by TBX5 gene mutations, located on the 12q24.1. This disorder displaying genetic heterogeneity is characterized by upper limb anomalies and congenital heart defects, with arrhythmia or not. These patients have asymmetry of upper limb lesion and normal intelligence. The incidence is 50% in the next generation. It is a monogenic disease with 10-50 times higher incidence compared with congenital heart disease caused by multiple genetic disorders. With the opening of the two-child policy in China, the birthrate of HOS may be increasing. At present, there are only a few ultrasound reports and fewer cases from the department of obstetrics and gynecology about HOS in China.

Objective
To improve the recognition of Holt-Oram syndrome, we probed into the clinical characters of this syndrome and reviewed the related literatures.

Methods
This study collected cases during August 2016 to December 2017, including heart and upper extremity deformities. According to clinical diagnostic criteria: A: Familial HOS: (1) with the character of autosomal dominant inheritance (2) the most common malformation in cardiovascular are ventricular septal defect or atrial septal defect (3) the upper limb skeletal abnormalities are mainly about phalanges carpus or radial abnormality. The above two can be diagnosed. B: Non-Familial HOS: It is necessary to have both cardiovascular and upper extremity deformity.

Conclusion
Holt-Oram syndrome is rare but with high incidence in the next generation. Although with severe structural abnormalities, 75%-80% babies were still born, which could be easily misdiagnosed in prenatal. The study enhanced the ultrasound doctor’s and gynecologist’s awareness of Holt-Oram syndrome and improved the prenatal detection rate of ultrasound. It could provide the basis for prenatal consultation and reduce the birth of children with Holt-Oram syndrome effectively.