HYDRAMENCEPHALY WITH ANTERIOR ABDOMINAL WALL DEFECT- A CASE REPORT
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PRESENTATION OF CASE
A 21 year old primigravida delivered a stillborn female baby with weight of 2.6 kg in the Department of Obstetrics and Gynaecology, Tezpur Medical College & Hospital, Tezpur, Assam. There was no family history of any genetic or congenital anomalies. The patient didn't smoke and there was no history that suggested any exposure to toxins or other chemical agents. The patient did not attend the antenatal clinic regularly and had not undergone routine US scan, anomaly scan or growth scan of the foetus during pregnancy. The foetus of 35 weeks gestational age was sent to the Department of Anatomy for study of congenital anomalies after taking written consent from the parents. The foetus was immediately fixed with 10% formalin. The foetus was then examined for internal and external defects. On examination, there was almost complete absence of the cerebral hemispheres and it was replaced by a membranous sac filled with cerebrospinal fluid and necrotic debris covered by leptomeninges. There was associated anterior abdominal wall defect with evisceration of developing bowel loops and liver with no membrane covering them. The foetus was dissected to detect any underlying abnormality. On dissection the other abdominal viscera were found to be normal, but the left lung and rib cage was found to be absent.

On examination the foetus was found to have features of Hydranencephaly with anterior abdominal wall defect. The foetus presented with absence of the cerebral hemispheres and it was replaced by a membranous sac containing cerebrospinal fluid and necrotic debris. On examination one edge of the placenta was found to be attached to a portion of the membranous sac. There was however preservation of some part of the skull. Intestinal loops were found to protrude through the anterior abdominal wall defect. The foetus was found to have Hydranencephaly associated with Gastroschisis. Babies with Gastroschisis are born with portions of their small and large intestine exposed outside the body through a defect in the abdominal wall usually just to the right of a normal insertion of the umbilical cord into the body wall and there is no covering membrane or sac.

DEFERENTIAL DIAGNOSIS
1. Extreme hydrocephalus: Where there is accumulation of fluid in the brain cavity which usually causes increase pressure inside the skull.
2. Bilateral extra cerebral collection of fluid in the skull
3. Severe open lip Schizencephaly.
4. Porencephaly: A disorder of the central nervous system involving cerebral cysts or cavities in cortical brain tissue.

DISCUSSION
Hydranencephaly is a rare congenital abnormality affecting the cerebral mantle. In this condition the cerebral hemispheres are completely or almost completely absent and are replaced by a membranous sac filled with cerebrospinal fluid. It is an extremely rare abnormality occurring in less than 1 per 10,000 births worldwide. The incidence worldwide is 0.2% in infant autopsies. Hydranencephaly is not compatible with a prolonged life after birth. The etiopathogenesis of hydranencephaly is heterogeneous and several theories have been postulated for its occurrences. The most common aetiology described is the occlusion of the supraclinoid segment of the bilateral internal carotid arteries causing ischaemic degeneration of structures supplied by them. Other aetiologies include an extreme form of leucomalacia formed by confluence of multiple cystic cavities, diffuse hypoxic-ischaemic brain necrosis, thromboplastic material from a deceased co-twin and intrauterine infections e.g. congenital toxoplasmosis or other viral infections like adenovirus, cytomegalovirus, enterovirus, Ebstein Barr virus, herpes simplex virus and respiratory syncytial virus. Hydranencephaly can be diagnosed in-utero on ultrasound examination and an MRI. An intrauterine CT should be used to support the sonographic assessment.

A female dead foetus delivered spontaneously by vaginal delivery in the Department of Obstetrics and Gynaecology, Tezpur Medical College & Hospital, Assam, was sent to the Department of Anatomy for study of congenital anomalies after taking informed consent from the parents.

Gastroschisis and omphalocele are commonly described anterior wall defects. Gastroschisis is characterised by an intact umbilical cord and evisceration of bowel through a defect in the abdominal wall generally to the right of the cord with no membranous covering. Omphalocele is...
characterised by herniation of bowel, liver and other organs into the intact umbilical cord, the tissues being covered by a membrane.  

The incidence of gastrochisis ranges between 0.4 and 3 per 10,000 births and seems to be increasing whereas the incidence of omphalolele ranges between 1.5 and 3 per 10,000 births and is stable.  

Five aetiologies of Hydranencephaly have been described:  
1) Bilateral occlusion of the supraclinoid segment of the internal carotid arteries or of the middle cerebral arteries.  
2) An extreme form of leukomalacia formed by confluence of multiple cystic cavities.  
3) Diffuse hypoxic-ischemic brain necrosis. Foetal hypoxia due to maternal exposure to carbon monoxide or butane gas may result in massive tissue necrosis. Subsequent cavitation and resorption of necrotized tissue creates the characteristic findings.  
4) Infection - necrotizing vasculitis or local destruction of the brain tissue: congenital toxoplasmosis, cytomegalovirus and Herpes simplex infections (HSV) have been associated with multiple cases of hydranencephaly. There is an increased incidence of spontaneous abortion following a maternal Herpes infection in early gestation. Foetal infections with HSV and toxoplasmosis are frequently associated with central nervous system and ocular anomalies later in gestation.  
5) Thromboplastic material from a deceased co-twin: monochorionic twins have presented with a variety of cerebral lesions. Lesions in the recipient twin result from emboli or thromboplastic material originating from the macerated co-twin. Coincident blood pressure instability and episodes of severe hypotension may lead to brain and visceral lesions in the recipient twin.  

Exact cause of gastrochisis is uncertain, but various causes have been proposed including ischaemic injury to the anterior abdominal wall (absence of the right omphalomesenteric artery) and weakness of the wall caused by involution of right umbilical vein. Gastrochisis has a very strong association with young maternal age (< 20 years). In addition, Gastrochisis has been linked to maternal exposure to cigarette smoking, illicit drugs, vasoactive over the counter drugs (such as pseudoephedrine) and environmental toxins.  

DISCUSSION OF MANAGEMENT

Hydranencephaly has a poor prognosis as majority of brainstem functions are missing. Affected newborns can die at birth but most infants die within the first year of their life. If a child does survive, they will be severely handicapped.  

Counselling the parents regarding the poor prognosis and the potential management options is advised to help them prepare for the potential outcome. For the child with HE who survives there is debate as to whether or not to perform any surgical treatment considering the severe brain impairment. The surgical treatment consists of ventriculoperitoneal shunt, which drains the fluid, reducing the cerebral tension and the progressive increase of the cerebral volume.  

Abdominal wall defects are often diagnosed by prenatal ultrasound done for routine screening or for obstetric indications such as evaluating an elevated alpha-fetoprotein of serum (AFP). In Gastrochisis, maternal serum AFP is markedly elevated, with an average elevation of nine multiples of mean (MoM). Prenatal ultrasound could potentially identify the overwhelming majority of abdominal wall defects and accurately distinguish omphalolele from Gastrochisis. Improved understanding of Gastrochisis, its early diagnosis by prenatal ultrasound, safe delivery of the foetus with a ventral wall defect, advanced surgical techniques for its correction and intensive care management of neonates reduces the morbidity and mortality. Mainstay of treatment of such anterior wall defects is to reduce herniated viscera into the abdomen and to close with fascia and skin to create a solid abdominal wall with relatively normal umbilicus while minimising risk to the baby. Patients with Gastrochisis have good survival rate of 90 to 95% provided they do not have catastrophic bowel loop sepsis and long-term complications of short bowel syndrome.
Figure 3. Absence of cerebral hemispheres which was replaced by a membranous sac containing cerebrospinal fluid and necrotic debris.

Figure 4. Presence of right lung and rib cage, but the left lung and the rib cage are absent.

FINAL DIAGNOSIS
Hydranencephaly with Gastroschisis.

REFERENCES