Case Report

Chaos created by CHAOS!! : A Case Report

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Abstract

A 22 years old unbooked primigravida with 18 weeks amenorrhea came to the department of obstetrics and gynecology at Sri Aurobindo hospital for a routine checkup. On her sonography, the fetus was found to have multiple congenital malformations. The post abortus fetus, on autopsy, turned out to be **one of the extremely rare cases of CHAOS. CHAOS** (Congenital High Airway obstruction Syndrome) is a rare and commonly fatal abnormality pertaining to innate obstruction of the fetal airway. Most authors postulate that this malformation is related to a cessation of the embryological development of the Sixth brachial arch at different gestational junctures. In our case of CHAOS, a confirmation of the preliminary diagnosis, with location of the precise site, level and degree of obstruction was attained with the help of an autopsy. Foetal autopsies should be encouraged as they symbiotically benefit the parents and the clinicians to a more clear and precise diagnosis.

Key Words: CHAOS, Foetal autopsy, congenital malformation, Embryological Development

Introduction:

A foetal autopsy is one that is performed on a dead born or still born foetus. Among the foetal autopsies performed in our department, we have come across several rare cases like, Meckel Gruber syndrome, Prune Belly syndrome, umbilical stricture and meconium peritonitis.

CHAOS (Congenital High Airway obstruction Syndrome) is a rare and commonly fatal abnormality pertaining to innate obstruction of the fetal airway. The obstruction may be partial or complete. [1]

A. C. Vidaeff et al [2] conducted a literature search between 1965 and January 2006 using the Pubmed bibliographic database. Their search yielded 36 prenatally diagnosed cases of upper airway obstruction.

Our Radio-diagnosis department conducted a literature research, also using Pub med Bibliographic database, between 2007 and December 2012. The key words used for the search were CHAOS, laryngeal stenosis, laryngeal atresia, ultrasonography and prenatal.

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DOR: 25.02.2014 DOA: 20.05.2014

The research concluded that there were **three** prenatally diagnosed cases of laryngeal atresia in this stretch. Laryngeal atresia is the most common etiology of CHAOS.

Other causes comprise of laryngeal or tracheal webs, subglottic stenosis or atresia, laryngeal cyst, tracheal atresia, or tracheal agenesis. In majority of the cases, the subglottic portion of the airway (laryngeal) or proximal trachea is atretic or stenotic.

A thick web may also be noticed, obstructing the proximal airway. [3]

The definitive cause is not known. Most authors postulate that this malformation is related to a cessation of the embryological development of the Sixth brachial arch at different gestational junctures.

Genetic, sporadic, and vascular causes have also been suggested. [4] The obstructed airway causes a diminished clearance of fluid produced by the fetal lungs which in turn causes an increase in intra-tracheal pressure that result in lung hyper-expansion and abnormal development. [3, 5]

The hyper-expanded lungs then compress the heart and inferior vena cava, reducing the venous return and precipitating intrauterine fetal congestive heart failure and non-immune hydrops. [3, 5, 6]

Case History:

A 22 yrs old unbooked primigravida with 18 weeks amenorrhea came to the Department of Obstetrics and gynecology of Sri Aurobindo hospital for a routine check-up. On her sonography, the fetus presented with multiple

congenital malformations. The patient was advised Medical termination of pregnancy and the post abortus fetus was sent for autopsy.

Autopsy Findings:

Before performing the autopsy a written informed consent was taken from the parents in their vernacular language. A team of doctors including Forensic experts, a pathologist and a radiologist, was formed.

External Examination:

The foetus presented with **Iniencephaly and anencephaly with rachischisis**. The **brainstem and spinal cord were seen exposed and cerebrum and cerebellum was absent.** (Fig.1) The foetus weighed 160 gm with a crown rump length of 8 cm, rump heel length of 7 cm, chest circumference of 14 cm and abdominal circumference of 14 cm.

Eyes, ears, nose, mouth, anus, external genitalia, placenta were all grossly normal. Skin was translucent and edematous.

Umbilical cord showed two umbilical arteries and a single umbilical vein. The pleural surface was grossly normal and fluid was present in both pleural cavities. The peritoneal surface was grossly normal. Fluid was present in the peritoneal cavity. The diaphragm was everted. The liver weighed 6.6gm. The spleen weighed 0.1 gm.

Stomach, small intestine, large intestine were all grossly normal. The pericardial surface and pericardial cavity were grossly normal. The oesophagus was patent and no trachea-esophageal fistula was noted.

On examination of the respiratory system, **larynx was completely blocked on probing with guide wire (4mm below the epiglottis) with post stenotic dilation of trachea and bronchus.** (Fig. 2, 3)

The left and right **lungs weighed 7.4 g and 7.8 g respectively** and **were enlarged in size with rib notching seen on right lung up to 11th rib. The heart was compressed and weighed 0.2gm**.

The great vessels arising from the heart and arising from the aortic arch did so in the normal position. Myocardium, mural and valvular endocardium; Foramen ovale; Ductus arteriosus; all the valves; Coronary ostia and coronary sinus; thoracic and abdominal aorta were grossly normal.

The gastrointestinal system, genitourinary system, hematopoietic system, musculoskeletal system were all grossly normal.

Hence, from the positive findings in the given foetus, the final diagnosis was given as **"Iniencephaly and Anencephaly with CHAOS."** CHAOS presents with massively enlarged lungs and a small compressed heart, as can be seen below in a comparative analysis of the given foetus with a normogram, of different organs at the same gestational age.

Discussion:

CHAOS is a rare congenital abnormality which in majority of the cases has a lethal outcome. It presents with a range of abnormalities, as obstruction or severe narrowing of upper airway which maybe in the form of laryngeal atresia, laryngeal web, subglottic stenosis, laryngeal cyst or tracheal obstruction; massively enlarged lungs; a flattened/everted diaphragm; a dilated trachea-bronchial tree.

Other abnormalities are a compressed small heart; ascites/pleural effusion, hydrops in utero; any tracheo–oesophageal fistula and may also present with other structural abnormalities for example laryngeal atresia with cryptopthalmos, renal agenesis, syndactyly, genital abnormalities which constitute the **FRASER syndrome.** [1-3, 5- 9]

We are reporting a case of CHAOS which presented with a primary abnormality of complete intrinsic foetal airway obstruction due to laryngeal atresia and also had associated findings of inencephaly, a term that was derived from the **Greek word "inion"** for nape of the neck. It is a rare type of cephalic disorder that was first described by **Etienne Geoffroy Saint-Hilaire in 1836.** [10]

Those afflicted with the disorder share three common characteristics: [10] Occipital bone defect, partial or total absence (rachischisis) of Cervico-thoracic vertebrae and fixed fetal head retroflexion.

So what is the need for Foetal Autopsy?

The usefulness of foetal autopsy cannot be definitively calibrated. For instance, lethal skeletal dysplasia is a condition where an autopsy can metamorphose the final diagnosis from Jeune's syndrome, (which has a high recurrence risk), to thanatophoric dysplasia (which presents with low recurrence risk), where prenatally only a presumptive diagnosis can be made. [12, 14]

Similarly, in renal cystic disease histological examination helps to differentiate between cystic renal dysplasia (recurrence risk 3%) and infantile polycystic kidney disease (recurrence risk 25%), which may otherwise be missed on ultrasonography scan owing to scarcity of amniotic fluid.

In our case of CHAOS, a confirmation of the preliminary diagnosis, with location of the precise site, level and degree of obstruction can only be attained with the help of an autopsy.

MTP Act 1971 allows termination of pregnancy if there is substantial risk that if the child were born it would suffer from such physical or mental abnormalities as to be seriously disabled. Some parents, in accordance with the act, may request termination of pregnancy, on the basis of prenatal investigations.

It imply that the unborn fetus will almost certainly suffer from a anomaly with a lethal outcome, for example, inencephaly, or one which would compromise the lifestyle of the baby with its persistent morbidity, for example Down's syndrome. Sometimes routine USG misses condition like umbilical stricture which results in sudden second trimester intrauterine deaths and is detected only on autopsy. [13]

An autopsy examination will not only provide parents with a confirmation of a doubtful prenatal prediction but also help ascertain the implications of a similar condition in future pregnancies. An autopsy leads to the refinement in the risk of recurrence, as was evident from a retrospective study done on 57258 deliveries, which showed that, when the final prenatal diagnosis was made by ultrasound scan, in 27% of the cases the autopsy added information that led to a decrement in the risk of recurrence. [14]

In cases where a definitive diagnosis cannot be made even by autopsy, genetic analysis and chromosomal studies is carried out to reach an accurate conclusion, and when a prenatal diagnosis is made then an autopsy may reveal some additional malformation which may help in forming a syndrome and addressing the same in successional pregnancy tests.

On a broader spectrum, a foetal autopsy not only improves the diagnostic quality, but is also valuable for the field of research, teaching and in discovering evolving anomalies.

Conclusion:

The autopsy findings of the given foetus were consistent with Iniencephaly and Anencephaly with CHAOS. Foetal autopsies should be encouraged as they symbiotically benefit the parents and the clinicians to a more clear and precise diagnosis.

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Fig.1: Iniencephaly and Anencephaly with Rachischisis

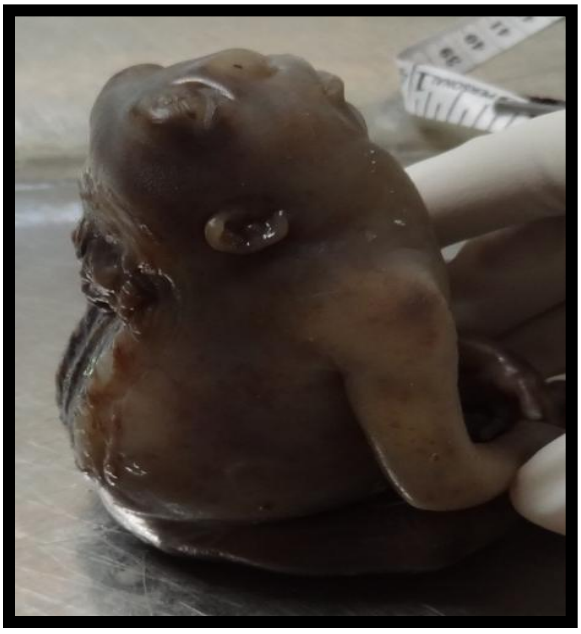

Fig 2: Eliciting Stenosis on Probing With Guide Wire

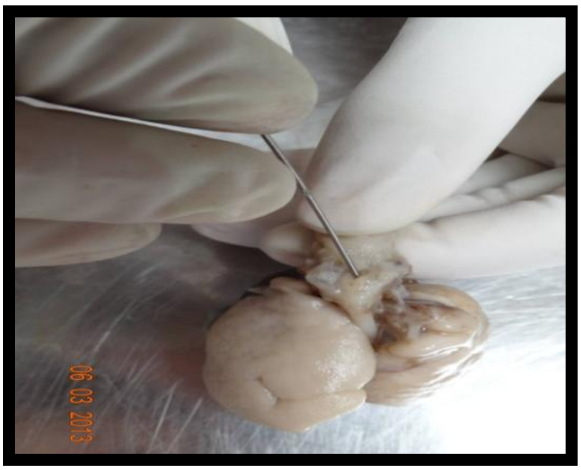

Fig 3: Complete Blockage of Larynx 4 Mm Below Epiglottis

Tab 1: Normogram of Different Organs at 18 Weeks of Gestation

in the given fetus:
age- 18 wks; body wt- 160 gm; lungs- 7.4 gm (L) +7.8 gm (R); heart- 0.2 gm

Age (wk)	Body	Brain	Thymus	Lungs	Heart
12	20.9 ± 6.6	3.20 ± 1.44	0.01 ± 0.01	0.50 ± 0.28	0.15 ± 0.0
13	31.2 ± 10.1	5.19 ± 1.95	0.03 ± 0.01	1.08 ± 0.45	0.20 ± 0.0
14	49.1 ± 14.5	8.14 ± 2.58	0.05 ± 0.02	1.79 ± 0.67	0.31 ± 0.1
15	74.7 ± 19.8	12.0 ± 3.3	0.09 ± 0.04	2.64 ± 0.92	0.50 ± 0.1
16	108 ± 26	16.9 ± 4.2	0.14 ± 0.06	3.61 ± 1.21	0.76 ± 0.2
17	149 ± 33	22.8 ± 5.2	0.20 ± 0.08	4.70 ± 1.55	1.10 ± 0.3
18	197 ± 42	29.7 ± 6.3	0.28 ± 0.1	5.92 ± 1.92	1.50 ± 0.4
19	255 ± 51	37.2 ± 7.6	0.41 ± 0.17	7.30 ± 2.34	1.88 ± 0.4
20	319 ± 61	45.7 ± 8.9	0.54 ± 0.23	8.84 ± 2.80	2.41 ± 0.5

Age- 18 wks; Liver- 6.6 gm; spleen- 0.1 gm; kidneys- 0.5 + 0.5 gm

Age (wk)	Liver	Spleen	Adrenals	Pancreas	Kidneys
12	1.01 ± 0.38	0.01 ± 0.01	0.10 ± 0.03	—	0.16 ± 0.04
13	1.38 ± 0.57	0.01 ± 0.01	0.15 ± 0.05	—	0.22 ± 0.07
14	2.18 ± 0.84	0.03 ± 0.02	0.23 ± 0.08	—	0.36 ± 0.13
15	3.41 ± 1.18	0.05 ± 0.03	0.33 ± 0.12	—	0.59 ± 0.19
16	5.06 ± 1.60	0.09 ± 0.05	0.47 ± 0.16	—	0.90 ± 0.28
17	7.14 ± 2.10	0.15 ± 0.07	0.64 ± 0.22	—	1.30 ± 0.39
18	9.65 ± 2.6	0.21 ± 0.10	0.84 ± 0.30	—	1.79 ± 0.51
19	12.8 ± 3.3	0.30 ± 0.14	1.03 ± 0.34	—	2.36 ± 0.65
20	16.5 ± 4.0	0.41 ± 0.18	1.29 ± 0.41	0.50 ± 0.14	3.00 ± 0.81

Quantitative Standards for Fetal and Neonatal Autopsy
John G. Archie et al