A Syndrome of Giant Omphalocele, Hypoplastic Left Chest Left Congenital Diaphragmatic Hernia, Cryptorchidism, Vertebral Deformities etc. – A Case Report

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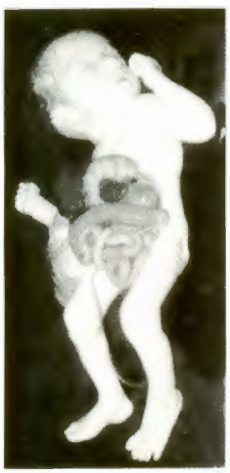
A 25 years old primigravida was admitted in a private nursing home on 6th March, 2000 with 9 months amenorrhoea and labour pain since 4 hrs. There was no h/o infection, exposure to radiation, ingestion of drugs etc in antenatal period. There was no family history of any congenital anomalies. Her antenatal period was uneventful but antenatal check up was irregular & ultrasonographic examination has not been done inspite of repeated advice. On examination nails were pink, pulse rate was 84/min, regular, BP 130/82 mmHg with mild pedal oedema. Abdominal examination revealed gravid uterus of 36 wks size, transverse lie of fetus, mild hydramnios & mild uterine contractions. FHS was audible & FHR was 148/min, cervix was 3 cms dilated. Emergency LSCS was done for transverse lie of fetus on same day. Post operative period was uneventful & she was discharged on 8th post operative day.

Examination of the baby revealed a male baby of 2.2 kgs with multiple congenital anomalies. Umbilical cord was very short & only 20 cms long with normal placenta & umbilical vessels. A giant omphalocele that contained most of the abdominal viscera including whole of liver, spleen as well as the entire intestinal tract (Fig. 1). The peritoneal cavity was under developed & very small.

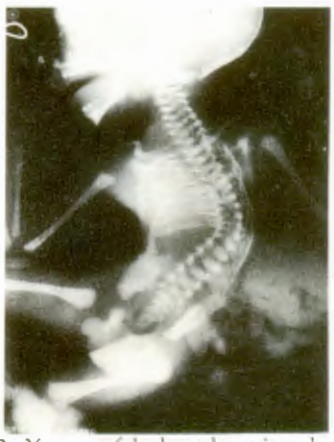


Photograph 1 Shows margin of omphalocele, intestines, liver, spleen, diaphragmatic hernia, hypoplastic left chest etc.

The omphalocele sac ruptured accidentally during operation. The length of intestines seems to be normal except malrotation. Severe hypoplasia of chest wall, ribs, pectoral muscles on left side were noticed. Costal cartilages on left side also absent & left nipple displaced laterally (Photograph. 1). Left congenital diaphragmatic hernia of 3x 2 cms without sac, severe degree of hypoplasia of left lung and dextrocardia were also present. Scoliosis at thoracic region towards right side & cryptorchidism were also noticed. Both kidneys, ureters & urinary bladder were normal.

Baby survived only 15 min. after birth & died due to respiratory failure. Post mortem x-ray of whole baby revealed hypoplastic left chest, dextrocardia, scoliosis towards right side, multiple thoracic vertebral deformities & soft tissue shadow of omphalocele (Photograph. 2).

Giant omphalocele containing entire intestines, liver & spleen with congenital diaphragmatic hernia have been reported in literature. Anterior abdominal wall defect as giant omphalocele with severe hypoplasia of chest wall, thoracic cavity, pectoral muscles, diaphragm & lung on left side as well as dextrocardia, cryptorchidism, scoliosis, vertebral deformities etc. in combination is a rare occurence; hence this case is being presented.



Photograph 2: X-ray of baby showing hypoplastic left chest, scoliosis thoracic region towards Rt. side, thoracic vertebral deformities, dextrocardia, soft tissue shadow of omphalocele etc.