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An unusual fetus with complete absence of thoracic, lumbar and sacral vertebrae, bilateral renal agenesis, VSD, meningomyelocele, imperforate anus, and teratoma

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We present a 30 week old male fetus who had a very interesting malformation complex which can not be explained by teratogenic or hereditary diseases. The aim of this paper is to discuss this complicated entity and compare it with other reported cases.
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Keywords: Agenesis of vertebrae and ribs, renal agenesis, imperforate anus, meningomyelocele, caudal regression sequence, single umbilical artery, absence of the left diaphragm

CASE REPORT

The 30-week-old male fetus was the first child of first cousins. The mother was 18 years and the father was 22 years old. At 30 weeks gestation ultrasonography showed severe anhydramnios, generalized ascites, short limbs, ventriculomegaly and approximately 5 weeks of intrauterine growth retardation.

There was no history of exposure to any teratogenic factors or radiation during the pregnancy and no history of maternal diabetes. Delivery was induced and the fetus was stillborn.

Postmortem blood chromosome analysis was not taken and culture of postmortem tissue cultures was unsuccessful.

AUTOPSY FINDINGS

The weight was 1850 g. The length from head to the end of the cystic mass was 32 cm. The circumferences of head, chest and abdomen were 29 cm, 12.5 cm and 26.5 cm respectively. There were Potter facies, short contracted limbs, club foot, imperforate anus and a huge cervico-thoraco-lumbo-sacrococcygeal cystic mass (Figure 1a and b). The size of the cystic mass was $17 \times 10 \times 6$ cm and extended along the back of the fetus and ended 2 cm below the neck. It contained bloody serous fluid. The fetus had a single umblical artery. There were male external genitalia. The chest wall was formed by only soft tissue. The left side of the diaphragm was absent. The left liver lobe protruded into the thoracic cavity, the left lung and heart were pushed to the right side. A Ventriculo-Septal defect (VSD), complete ileal stenosis, a blind ended large bowel, and anal atresia were also observed. The adrenal glands were found in their normal position, however the kidneys were missing on both sides (bilateral renal agenesis). The bladder was hypoplastic. There was another mass in the retroperitoneal region sized as $3 \times 2 \times 1.5$ cm, located on the right side of the aortic bifurcation. The right gonad was localized in the right pelvic region and its size was 1×1 mm but the left

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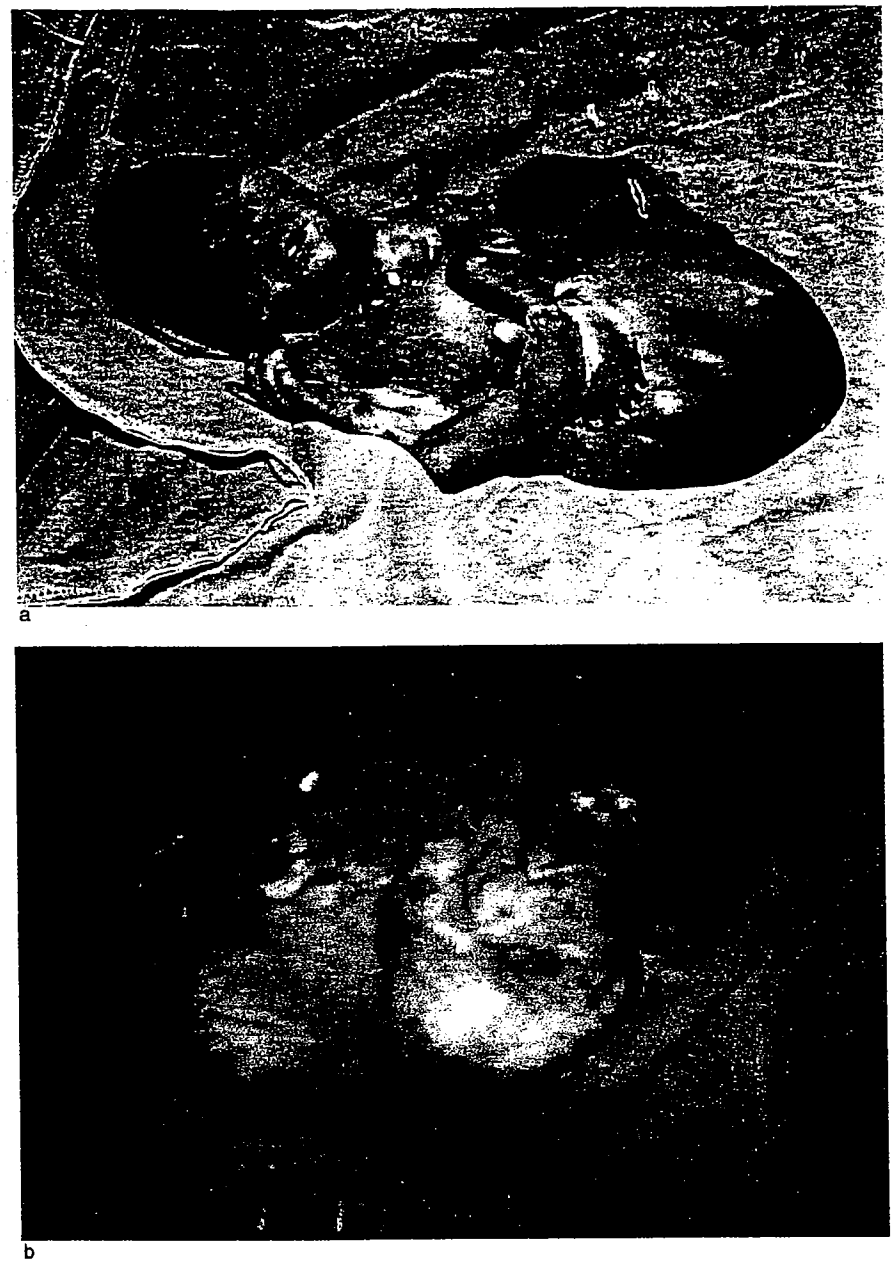

FIGURE 1. (a) The general appearance of the malformed fetus characterized by a huge cystic mass, short and contracted extremities and Potter facies. (b) Lateral view of the fetus. Cystic mass is extending from sacral region to the neck

gonad was absent. Enlargement of lateral ventricles was observed in the brain. Postmortem X-ray examination showed normal cervical vertebrae and total absence of thoracic, lumbar and sacral vertebrae, and ribs (Figure 2).

Microscopic findings; the wall of the cystic mass contained neural and glial tissue which are typical for meningomyelocele. The second mass located on the right side of the aortic bifurcation showed features of a teratoma. The lesion was made up of intermingled areas of striated muscle, adipose tissue, neurons, nerve bundles, renal tissue composed of glomerular, tubular structures, blastemal elements, bone and cartilage (Figure 3). The right gonad showed features of an immature testis. The other organs were unremarkable.

DISCUSSION

The present case had absence of thoracic, lumbar, and sacral vertebrae, ribs and left side of the diaphragm, bilateral renal agenesis, VSD, imperforate anus associated with high level anal atresia, ileal stenosis, a

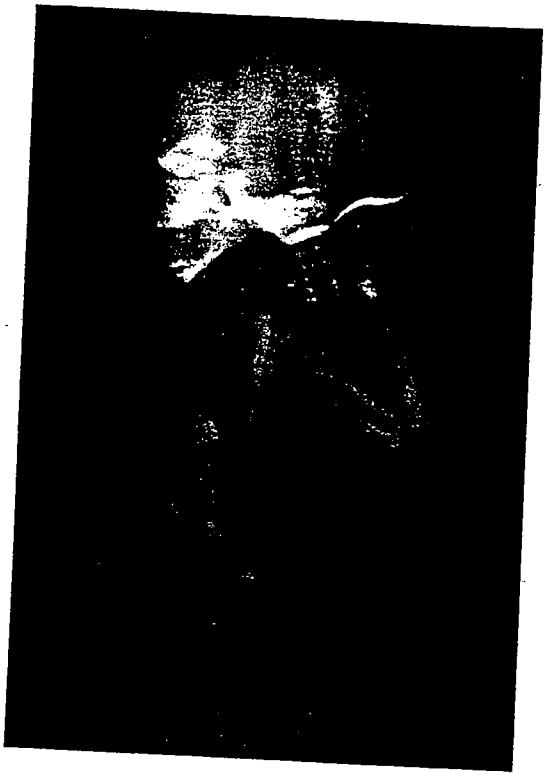

FIGURE 2. The postmortem X-ray of the fetus. Only cervical vertebrae were observed, thoracic, lumbar, sacral vertebrae and ribs were absent.

cystic cervico-thoraco-lumbo-sacrococygeal meningomyelocele and an additional retroperitoneal teratoma. Potter and Craig (1975) reported a case with absence of lower thoracic and upper lumbar vertebrae such as in our case. The present case is more severe as there are only cervical vertebrae. Absence of lumbar and sacral vertebrae are most commonly observed in diabetic mothers (Banta and Nichols, 1969). However Potter reported two infants of non-diabetic mothers with lack of all of the lower dorsal and most of the lumbar and sacral vertebrae (Potter and Craig, 1975).

Rossi *et al.* (1995) described a 32-week-old male fetus who had extreme hypotrophy of the lower body pole, extensive hypoplasia of the spinal column and multiple anomalies of abdominal organs similar to the present case and suggested an extreme variant of the caudal regression sequence Most of the findings of our case are similar to that case such as severe costovertebral and lower body pole hypotrophy and multiple internal anomalies (single umblical artery, renal agenesis, imperforate anus, diaphragmatic hernia, spinal and rib defects). Caudal regression syndrome is characterized by incomplete development of the sacrum and/or lumbar vertebrae, absence of the body of the sacrum leading to flattening and dimpling of the buttocks, shortening of the intergluteal cleft, and incontinence of urine and faeces. Renal agenesis, imperforate anus,

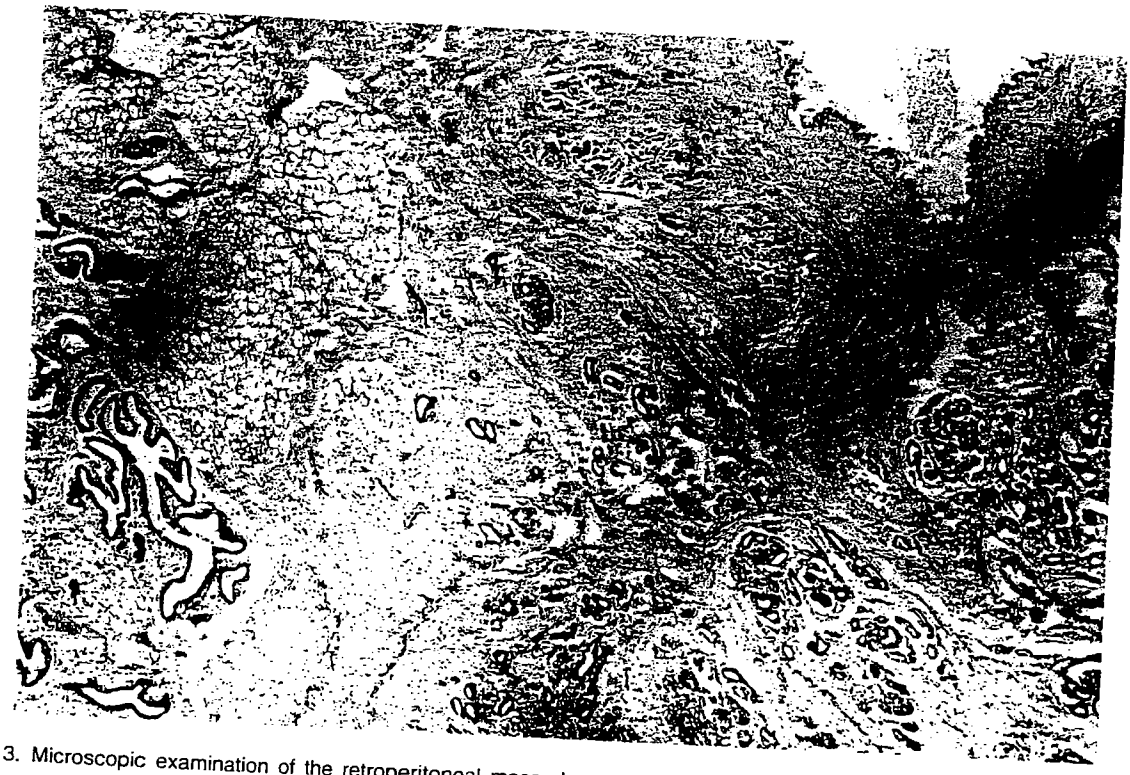

FIGURE 3. Microscopic examination of the retroperitoneal mass showed a teratoma. Heterogeneous collection of cystic mucinous epithelium, cartilage and renal tissue is seen here (H.E. ×40)

cleft lip, cleft palate, microcephaly and meningomyelocele are occasionally observed (Jones, 1997). Passarge and Lenz (1966) reported the association between caudal regression syndrome and maternal diabetes. Gürakan *et al.* (1996) also reported an association between diabetes and sirenomelia. Animal studies provide support for maternal diabetes as a cause of caudal regression syndrome (Gale, 1991). Vascular obliteration has also been suggested as the cause of sirenomelia and associated defects (Stevenson *et al.*, 1986; Akbıyık *et al.*, 2000). In our case the thoracic and abdominal aorta was normal and there was also a teratoma, VSD and meningomyelocele.

Teratoma is the most common tumour of the newborn (Gonzales-Crussi, 1982; Isaacs 1985). In our case, there was a simultaneous presentation of sacrococcygeal meningomyelocele and teratoma. Meningomyelocele and teratoma have been reported previously (Reid and Mickle, 1985; Sherer *et al.*, 1997; Koen *et al.*, 1998; Özer and Yüceer, 1999) but other malformations observed in our case have not been associated.

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