P04.08: Serpentine-like syndrome in a fetus presenting with abnormal median diaphragmatic hernia

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Introduction: Serpentine-like syndrome was first described in 2008. Since then, 11 cases have been reported in literature. The syndrome is characterized by brachyesophagus, secondary intrathoracic stomach and vertebral and spinal anomalies. Further associated anomalies such as agenesis, malposition or herniation of abdominal organs have been described.

Case report: We report on a 28-year-old G1 who was first seen in 13+5 weeks of gestation with a cystic structure in the posterior mediastinum without further fetal abnormalities at this time. The karyogram revealed a normal female karyotype. In the further course of pregnancy the heart was anteponed with the stomach posterior on the right side, and intestinal loops in the posterodorsal mediastinum. The LHR in 26+5 SSW was 53%, the fetal liver was intraabdominal at all times. In the third trimester a significant polyhydramnion developed which was treated with amniotic drainage in 35+3 weeks of pregnancy due to maternal symptoms.

Delivery was performed as a secondary cesarian in 37+1 weeks. On the 2nd day surgery was performed and revealed a large central diaphragmatic defect with an intrathoracic position of the stomach and a severe brachyesophagus (approx. 4 cm). Parts of the duodenum and pancreas were found in the mediastinum. The duodenum could be repositioned. Stomach and pancreas could not be relocated intraabdominally in first surgery. An MRI-scan of the spine was performed on the 6th day and showed a spinal cord compression through atlas and axis with narrowing of the spinal canal to 5 mm. A laminectomy and decompression of the cervical myelon was performed neurosurgically on the 17th day.

Conclusion: In patients with congenital intrathoracic stomach a “serpentine-like syndrome” should be considered as differential diagnosis and appropriate examinations should be performed. In our case the atypical intraoperative findings and the literature search were the key to the diagnosis of this very rare disease. Thus, the cervical anatomy could be detected early and further complications could be avoided.