A case of Klippel-Feil Syndrome

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Objective

Klippel- Feil Syndrome is a very rare disease, which is characterized by three symptoms: short neck, limitation of head and neck movements and low posterior hairline. The disorder is estimated to occur in 1 in 40, 000 to 42, 000 newborns worldwide. Females are affected slightly more often than males. The skeletal disorder is primarily characterized by abnormal union or fusion of two or more bones of the vertebrae within the cervical vertebrae. It is believed to result from faulty segmentation along the embryo's developing axis during the 3rd to 8th weeks of gestation. Klippel-Feil syndrome can occur with other syndromes such as fetal alcohol syndrome, Goldenhar syndrome, and abnormalities of the arms or legs.

Methods

The aim of this study was to present a case report of the fetus with the diagnosis of Klippel- Feil Syndrome.

Results

A pregnant woman was admitted to the Outpatient Clinic of Diagnosis and Therapy of Fetal Defects, Department of Obstetrics in the 12th week of pregnancy, diagnosed with increased nuchal transluciency in the fetus. Therefore genetic amniocentesis was performed. In result a normal male karyotype was recognized. The pregnancy was not complicated by other diseases. There were no effects of environmental factors found on the current pregnancy. There were abnormalities within cervical spine identified in the 20th week of pregnancy. Morover, irregularities were found within the cervical spine and deep, around considerable with shortening it and irregularities in the range of the chest. The pregnancy was finished in the 40th week through elective caesarean section and a male newborn weighing 2600g was born with 6 points according to Apgar score. Due to the respiratory failure, after the birth the newborn stayed intubated. Underdevelopement of the lungs was recognized and in consequence the inborn pneumonia was complicated with artificial pneumothorax. What's more, because of the disorders of etching, the infant was submitted to the Department of Childrens Intensive Therapy in Bydgoszcz.

Conclusion

It is significant to properly recognize the type of malformation within the bone structure, in order to prenatally determine the appropriate clinic, in which the pregnancy should be finished. Key considerations in the emergency management of such patients include radiographic evaluation for hypermobile cervical segments, the anticipation of difficult tracheal intubation, and proper follow-up and referral because of the progression of the skeletal pathology.