**Abstract**

**Objective:** We discussed a rare case of spinal muscular atrophy (SMA) type 0, was followed with prenatal diagnosis of intrauterine growth restriction (IUGR), five times of cord around neck detected during cesarean section, in the highlights of the literature.

**Case:** A pregnant woman who was followed with the diagnosis of IUGR during 32. gestational week. In the 38. gestational week; cord around neck for five times and a real knot were detected in the cesarean section. Difficulty in swallowing and absorbing, also significant respiratory muscle weakness were found at the follow up of the baby. SMA was suspected according to these symptoms. With the analyse of PCR RFLP (Polymerase chain reaction – restriction fragment length polymorphism) SMN 1 (Telomeric survival motor neuron gene) homozygot exon 7 and 8 deletions were found, SMA type 0 was diagnosed according to these findings.

**Conclusion:** During the 2. trimester of pregnancy, with a careful ultrasound examination, cord around neck can be successfully determined. It should be considered that cord around neck risk rate can be increased in the 3. trimester. Hypotonia, severe respiratory muscle weakness involving the infants to be a good antenatal and postnatal history, examination and molecular study will identify the rare cases of SMA. Genetic counseling should be given to the parents.

**Keywords:** Pregnancy, umbilical cord around neck, intrauterine growth retardation, spinal muscular atrophy.

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**Introduction**

The coiling of umbilical cord around fetus neck is seen in 23-33% of all pregnancies and it is generally evaluated as benign.[1,2] It is reported that this situation can be seen at a rate of 6% on 21st gestational week.[3] It is less often to observe coiling or knotting of cord around fetus body and extremities.[4,5]
One of the most frequently seen genetic diseases causing child death around the world is autosomal recessively inherited spinal muscular atrophy (SMA).\[6\] While SMA was grouped into three sub-groups according to their clinical aspects and beginning age in previous years, Dubowitz defined severe cases (previously classified as SMA Type 1) as SMA 0. SMA Type 0 is clinically characterized severe symmetrical muscle weakness and flaccidity seen much more in lower extremities than upper extremities after birth and affected much more in proximal extremity than distal. Patients frequently require resuscitation and ventilator support at delivery.\[6-9\]

SMA occurs as a result of SMN (survival of motor neuron) gene mutations on long side of 5th chromosome.\[6-10\] In this article, SMA Type 0 case which was followed up by IUGR diagnosis in prenatal period and found cord coiled 5 times around neck during cesarean was discussed within the light of literature since it is seen rarely.

**Case Report**

Thirty-four years old, G2 P1 pregnant applied to our clinic for routine pregnancy follow-up. Brother of the patient had cerebral palsy, but there was no kin marriage history of the pregnant. At her 1st trimester screening, free beta HCG was 28.3 ng/ mL (0.61 MoM) and PAPP- A was 0.83 mIU/mL (0.63 MoM). In the ultrasonographical examination of fetus at 11th-14th gestational week, it was found that nuchal translucency was 1.9 mm, a wave in ductus venous blood flow was positive, there was no regurgitation on tricuspid valve, and nasal bone was 1.6 mm. Anatomy and development of fetus was normal. Notches were detected on bilateral uterine arteries. Fixed Down syndrome risk was calculated as 1/8136.

In the ultrasonographical examination of fetus at 22nd gestational week, biparietal diameter (BPD) was found as 54 mm, head circumference (HC) as 201 mm, abdominal circumference (AC) as 166 mm, femur length (FL) as 39 mm, fetal heart beat rate as 138 beat/min., and a cord existence located on placenta anterior wall and on neck. It was seen that a wave in ductus venous was positive and there was no regurgitation on tricuspid valve. Notching was continuing on bilateral uterine arteries. Anatomy and development of fetal was observed as normal. 1st hour blood glucose was found as 121 on 50 g oral glucose tolerance test (OGTT). In the ultrasonographical examination at 32nd gestational week, fetal measurements were found -2 below standard deviation (SD) according to gestational week. Estimated fetal birth weight was 1,352 g, amniotic fluid volume was normal and the patient detected breech presentation was taken to follow-up by IUGR diagnosis. There was cord coiled around neck two times.

The pregnant was followed up by fetal Doppler and non-stress test (NST) due to IUGR diagnosis until 38th gestational week. Single alive 2,230 gr male baby with 1st minute Apgar score 5 and 5th week Apgar score 9 was delivered by cesarean from the 38-weeks-and-3-days patient through cesarean undergone and breech presentation indications. The existence of cord coiled five times around neck (Figure 1) and true knot (Figure 2) was detected during cesarean. In the umbilical vein blood gas taken during delivery, following values were found: pH 7.36, PCO2 38, PO2 46, hematocrit 52. Also following values were found in the umbilical artery blood gas: pH 7.31, PCO2 50, PO2 23, hematocrit 53. Postoperative cord length was measured as 105 cm.

With the analysis of PCR RFLP (Polymerase chain reaction – restriction fragment length polymorphism) SMN 1 (Telomeric survival motor neuron gene) homozygote exon 7 and 8 deletions were found, then SMA type 0 was diagnosed according to these findings.

Patient who was diagnosed as sepsis three times (first one was when only one-month-old) due to pneumonia has still being followed up in pediatric intense care unit by providing respiratory support.

**Discussion**

The most frequent location of cord coiling which appears beginning from first trimester in the
pregnancy is the fetal neck circumference.\[11\] It was reported that cord may coil around neck beginning from the gestational week where cord length reaches 4/5 of fetus length.\[12\] It was stated that though normal term umbilical cord length is 55-75 cm, it may sometimes be 300 cm and sometimes aplasia may occur.\[21\] Cord length is considered as associated with coiling number.\[13\] In our case, cord length was determined as 105 cm.

When fetus was examined in terms of presentation, it was shown that cord coiling was frequently observed in breech presentations.\[14\] Breech presentation in our case was found as compatible with the literature.

The highest coiling number reported in the literature is nine.\[15\] In our case, 5 times cord coiling around neck was detected. Cord length, gestational week, placenta localization, fetus weight, fetus mobility and amniotic fluid amount are the factors associated with cord coiling.\[12,16\] In the literature, there are some studies reporting that placenta being on anterior wall also affects cord coiling.\[17\]
In our case, the placenta was found as anterior wall located.

It was reported that increasing mobility of fetus at third trimester was effective on cord coiling.\(^{13}\) In our case which was SMA type 0, expectation of fetal mobility decrease was not found as associated with cord coil when it was seen in delivery that cord coiled five times around fetus neck.

Clappa et al. reported that coiling rate increased as gestational weeks advanced.\(^{13}\) In our case, one time cord coiling was first detected on 22nd gestational week. At 32nd gestational week, two times cord coiling was detected at neck. During the cesarean performed on 38th gestational week, five times cord coiling and one true knot were detected.

SMA is a frequently seen neurodegenerative disease where it progresses with the degeneration of anterior horn cells of spinal cord and inherited by autosomal recessive. The prevalence in western societies is 1/6,000 – 1/10,000 and the carrier rate of the disease varies between 1/40 and 1/80.\(^{18}\) SMA’s other types as autosomal dominant and inherited according to X were also defined.\(^{19}\) In patients who are diagnosed as SMA type 0, asphyxia and generalized weakness are observed in newborn period. Most of them require resuscitation and ventilation support. Bulbar muscles are weak as causing weak crying, weak sucking and swallowing, and aspiration as a result of secretion aggregation. There is frequently fasciculation on tongue.\(^{6-9}\) In our case, minority of intrauterine fetal movements could not be examined clearly during fetal ultrasonographical examination or anamnesis taken from the mother. However, findings such as hypotonia and weakness were clearly observed during newborn examination. Pneumonia and sepsis developed within the first month were the most significant indication of weakness including respiratory muscles. It was shown in some studies that some of the cases who were diagnosed as having SMA after delivery could be followed by fetal IUGR diagnosis.\(^{20}\) In our case, IUGR was also observed beginning from 32nd gestational week. The diagnosis was established at postnatal period as in similar studies in the literature.

When other fetal hypocinesia cases were investigated in the literature, it was seen that birth dystocia, short umbilical cord, polyhydramnios, craniofacial malformations, multiple joint contractures and skeletal anomalies displayed association.\(^{20}\) In our case, cesarean was preferred as delivery method due to the cesarean history. In our case, unlike the literature, short umbilical cord (actually long umbilical cord was observed), polyhydramnios, craniofacial malformations and skeletal anomaly were not detected.

In our SMA type-0 case where intrauterine fetal movements were expected to decrease, observing cord coiling five times around neck and true knot during cesarean was not found compatible with the literature. Though it is not frequent to see cord coiling five times around neck in SMA cases, measuring cord length as 105 cm was considered as it can be explained by breech presentation of the fetus and anterior located placenta. By a careful ultrasonographical examination performed beginning from 2nd trimester of the pregnancy, it can be detected that umbilical cord is coiled around fetus neck. It should be remembered that this rate increases in weeks approaching 3rd trimester. In our case, cord existence around neck was detected by ultrasonographical examinations performed as of 22nd gestational week.

As seen in our case, SMA type-0 is a disease which should be considered first in differential diagnosis in patients who develop severe weakness, hypotonia and respiratory failure at newborn period. Together with examination findings and molecular study, receiving well antenatal and postnatal histories in newborns that have severe weakness including respiratory muscles will enable to know rare SMA cases and to give required genetic consultancy to families.

**Conclusion**

By a careful ultrasonographical examination performed beginning from 2nd trimester of the pregnancy, it can be detected that umbilical cord is coiled around fetus neck. It should be remembered that this rate increases in weeks approaching 3rd trimester.

Together with examination findings and molecular study, receiving well antenatal and postnatal histories in newborns that have severe weakness including respiratory muscles will enable to know rare SMA cases and to give required genetic consultancy to families.
References


