CASE REPORT

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CONGENITAL KYPHOSIS AND LUMBOSACRAL MENINGOMYELOCELE: PRENATAL FINDINGS OF A RARE ASSOCIATION – CASE REPORT

ANDRIELLE MIOZZO SOARES[1], CAROLINA DE MOURA MAROLLI[1], LEONARDO PFEIFER RUBIN[2], VALENTINA STEFFENS BRACHT[1], MARCELLE KLEIN DRAGHETTI[1], JORGE ALBERTO BIANCHI TELLES[3]

ABSTRACT

INTRODUCTION: Congenital kyphosis is a condition rarely described, especially in fetuses in the prenatal period, and its association with meningomyelocele is even more unusual. This case report describes the prenatal findings of a fetus with congenital kyphosis and lumbosacral meningomyelocele, highlighting the importance of evaluation by fetal ultrasound and magnetic resonance imaging for an accurate diagnosis and appropriate treatment planning.

CASE REPORT: A 35-year-old woman, in her second pregnancy, was referred to the fetal medicine service due to findings suggestive of fetal spina bifida. Fetal ultrasound and magnetic resonance imaging exams revealed the presence of significant congenital kyphosis, segmental spinal dysgenesis, lumbosacral meningomyelocele, hydrocephalus, as well as agenesis of the corpus callosum and septum pellucidum. Based on the findings observed, it was planned to perform the delivery and surgery, which occurred immediately after birth.

TREATMENT: the treatment involved planning for delivery and immediate surgery to correct the meningomyelocele identified in the prenatal exams, aiming to improve the quality of life and prevent complications associated with the diagnosed conditions. The patient also underwent a ventriculoperitoneal shunt due to hydrocephalus.

DISCUSSION: the importance of joint evaluation through fetal ultrasound and magnetic resonance imaging for the accurate diagnosis of complex conditions, such as congenital kyphosis associated with meningomyelocele, was emphasized. Additionally, the clinical and surgical implications of these malformations were discussed, highlighting the need for a multidisciplinary approach to provide the best treatment for the patient.

CONCLUSION: the case report highlights the importance of detailed prenatal evaluation in cases of complex malformations such as congenital kyphosis associated with meningomyelocele. The use of fetal ultrasound and magnetic resonance imaging allowed for an accurate diagnosis, as well as appropriate planning for delivery and surgical treatment, demonstrating the relevance of these complementary exams in clinical practice.

KEYWORDS: CONGENITAL KYPHOSIS, MENINGOMYELOCELE, PRENATAL DIAGNOSIS, ULTRASOUND, MAGNETIC RESONANCE IMAGING.

INTRODUCTION

Congenital kyphosis is a condition characterized by a malformation involving the spine, specifically the vertebral bodies, which undergo abnormal growth and posterior convex angulation. This condition can occur in isolation or be associated with other malformations, such as meningomyelocele[1]. It is worth noting that there are few reports in the literature of congenital kyphosis diagnosed prenatally, showing the findings identified through imaging exams, such as fetal ultrasound (US) and, especially, magnetic resonance imaging (MRI)[2].

Therefore, our objective was to describe the prenatal findings of a fetus with congenital kyphosis associated with meningomyelocele, emphasizing the characteristics identified in imaging exams, as well as their potential applicability in clinical and surgical practice.

CASE REPORT

The patient was a 35-year-old woman in her second pregnancy, who was referred to the fetal medicine service due to a fetal ultrasound with findings suggestive of fetal spina bifida. A new examination performed at our hospital drew attention to the presence of the "banana" and "lemon" signs, as well as abnormal closure of the lumbosacral spine associated with significant kyphosis (measuring 61 degrees, according to angular evaluation based on the ossification centers of the vertebral column) (Figure 1A and B).

1. Universidade Federal de Ciências da Saúde de Porto Alegre (UFCSPA), Porto Alegre, RS.
2. Pontifícia Universidade Católica do Rio Grande do Sul (PUCRS), Porto Alegre, RS.
3. Hospital Materno Infantil Presidente Vargas (HMIPV), Porto Alegre, RS.

MAILING ADDRESS:
JORGE ALBERTO BIANCHI TELLES
Av. Independência, 661 Bairro Independência
Porto Alegre, RS, CEP: 90035-074
E-mail: telles383@gmail.com

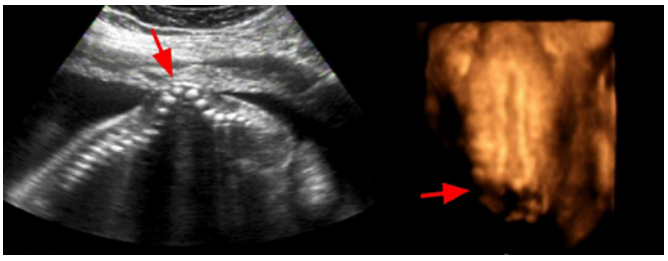

Figure 1. Two-dimensional and three-dimensional fetal ultrasound (A and B, respectively), performed at 24 weeks of gestation, showing congenital kyphosis and meningomyelocele (see the arrows).

Furthermore, there was a significant axial deviation of the lower limbs. Fetal MRI revealed segmental spinal dysgenesis and confirmed the presence of severe kyphosis. Additionally, posterior dysraphism associated with a cystic collection (suggestive of meningomyelocele), supratentorial hydrocephalus, partial parallelism of the lateral ventricles with enlarged occipital horns (suggestive findings of agenesis of the corpus callosum), and agenesis of the septum pellucidum were observed (Figure 2A). Additionally, there was a considerable reduction in the dimensions of the posterior fossa, with narrowing of the fourth ventricle and abnormal morphology of the cerebellum, which was inferiorly displaced and partially herniated into the vertebral canal. These findings were compatible with Arnold-Chiari malformation type II (Figure 2B).

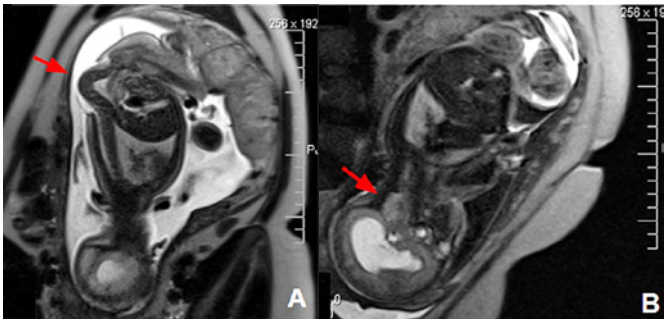

Figure 2. Fetal MRI images showing congenital kyphosis associated with meningomyelocele (A), as well as Arnold-Chiari malformation (B) (see the arrows).

The child was born via cesarean section, weighing 3,250g, measuring 45cm, with a head circumference of 37cm, and Apgar scores of 8 and 9 at the first and fifth minute, respectively. An important bony defect affecting the lumbar spine (kyphosis) was found, along with a meningomyelocele. Additionally, the suspicion of agenesis of the corpus callosum that arose during pregnancy was confirmed through MRI, as well as the presence of bilateral congenital clubfoot.

The patient underwent surgery to correct the meningomyelocele on the second day of life. A cerebral ultrasound, performed on the seventh day, showed supratentorial hydrocephalus, with the right lateral ventricle measuring 2.9cm and the left 3.7cm. There was also agenesis of the septum pellucidum, in addition to the previously described agenesis of the corpus callosum. Therefore, a ventriculoperitoneal shunt was performed at 16 days of life.

At three months, the child weighed 4,950g and was fed through a nasoenteric tube. It was also found that she had laryngomalacia. She was using a tracheostomy tube and receiving oxygen therapy via Ayre's T-piece, and she had an episode of convulsive crisis.

DISCUSSION

Defects in the formation of the vertebral column, such as the congenital kyphosis observed in our patient, can originate in different stages of embryonic development, but mainly occur during gastrulation and neurulation. During the gastrulation period (in the 2nd and 3rd weeks of gestation), the notochord originates and forms the basis for the development of the axial skeleton. In primary neurulation (in the 3rd and 4th weeks), the neural plate forms, which arises from the interaction between the notochord and the ectoderm, and its bending and folding to form the neural tube. This closes like a zipper, going in both directions[3].

Congenital kyphosis originates, therefore, from an anomaly in the formation of the notochord, which leads to a malformation of the vertebral bodies and, consequently, to different angles of the spine. The vertebral anomaly most commonly described in association with congenital kyphosis consists of hemivertebra, which usually affects more than one vertebral body (44%)[4].

Perhaps due to the proximity and timing of embryonic development (still in the first month of gestation), congenital kyphosis has been described in association with neural tube closure defects, as observed in our fetus, in up to 10 to 15% of cases. However, similar prenatal reports seem to be quite uncommon. Congenital kyphosis can also be associated with other conditions, including hemifacial microsomia/oculo-auriculo-vertebral spectrum (Goldenhar syndrome); Alagille, Jarcho-Levin, Joubert, and Gorlin syndromes, as well as Klippel-Feil sequence, VACTERL association, trisomy 18 (Edwards syndrome), and diabetic embryopathy[2].

Fetal ultrasound is an important prenatal tool capable of identifying not only structural abnormalities of the fetal spine but also congenital kyphosis. Furthermore, it is possible to diagnose associated alterations with great precision, such as neural tube closure defects[5], like the meningomyelocele identified in our case.

The evaluation of the fetal spine by ultrasound is commonly performed through the analysis of the ossification centers of the vertebrae, which are usually observed from the second trimester of pregnancy. They are located at two points in the neural arches and one in the vertebral body. Therefore, the main scanning planes used to assess the spine by ultrasound are the parasagittal, transverse, and coronal planes[5]. Most spinal defects are apparent between 20 and 22 weeks of pregnancy. In our case, the alterations were first visualized by ultrasound at 24 weeks, which is in accordance with what is described in the literature[6].

Through ultrasound, it is also possible to measure the degree of curvature of congenital kyphosis using assessment in the parasagittal plane. This method measures the angle formed between the line passing through the center of ossification of the most proximal vertebral body of the kyphosis with the line crossing the most distal center. Another method described in the literature for measuring the curvature of congenital kyphosis is by measuring the angle formed between perpendicular lines from the ends of the most cephalic and caudal vertebral bodies involved in the kyphosis. This method is analogous to the "Cobb angle," which is used to measure the degree of fetal scoliosis (see Figure 3). Ultrasound would also be a very reliable examination to determine the level of congenital kyphosis present in fetuses[7].

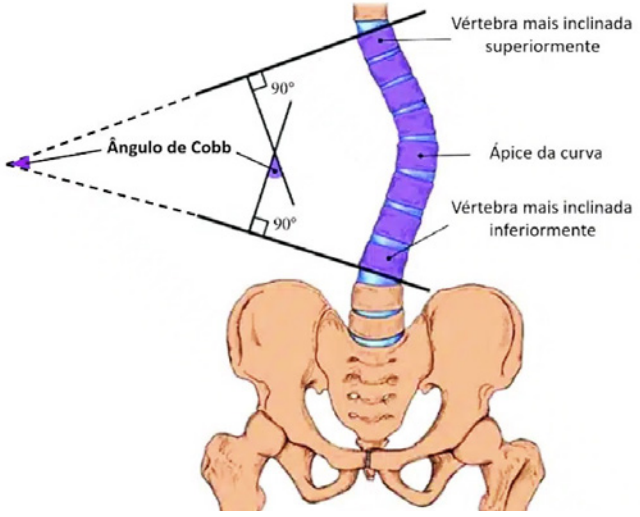

Figure 3. Illustrates the criteria for calculating the Cobb angle in scoliosis evaluation (Teixeira, R).

The findings observed on ultrasound suggesting the presence of meningomyelocele include the so-called "lemon sign" (in which there is overlapping of the fetal frontal bones) and the "banana sign" (resulting from the anterior curvature of the cerebellum towards the cisterna magna, which can lead to its obliteration). The ultrasound evaluation performed at our hospital was able to identify both signs, along with the defect in spinal cord closure at the lumbosacral level (meningomyelocele) associated with the spinal anomaly.

On the other hand, fetal MRI evaluation has been used as a complement to ultrasound in various situations. However, as mentioned earlier, there is almost no scientific literature describing prenatal reports of congenital kyphosis evaluated by fetal MRI2. In our case, this evaluation identified segmental spinal dysgenesis of the vertebral column, mainly affecting the lumbar segment, in addition to severe kyphosis associated with scoliosis.

In cases of neural tube closure defects, such as the meningomyelocele present in our patient, the importance of using MRI is already evident, as it is superior and complementary to ultrasound in evaluating certain abnormalities, such as involvement of nerve roots and the presence of Arnold-Chiari malformation observed in our fetus. The latter is characterized by herniation of the cerebellar vermis through the foramen magnum, caudally. It is best observed through sagittal sections obtained by fetal MRI. From this, it is also possible to determine the type of Arnold-Chiari malformation present, which in our fetus was type II, which is most commonly associated with meningomyelocele[8]. Other alterations in which this applicability exists include rhombencephalic hernia, cervicomedullary torsion, tectal beak, corpus callosum abnormalities, and heterotopias[3]. In our fetus, it was also possible to identify partial parallelism of the lateral ventricles, with greater volume increase of their occipital horns, findings indicative of agenesis of the corpus callosum. Additionally, agenesis of the septum pellucidum was noted.

Fetal MRI is also very important when considering surgery, including fetal surgery, to correct meningomyelocele, as it helps confirm the diagnosis and exclude the presence of associated anomalies. For all these reasons, we believe that the combined evaluation, through both ultrasound and MRI, is ideal.

Although there are conservative therapies, surgical correction remains the main treatment for congenital anomalies that significantly affect the spine, such as congenital kyphosis. Its main goal is to decompress neurological elements and correct the malformation. The surgical approach to be adopted varies according to the severity of the defect[9,10]. In our case, the surgical procedure was performed on the second day of life. Additionally, a ventriculoperitoneal shunt had to be subsequently performed due to hydrocephalus secondary to meningomyelocele.

Regarding prognosis, there is a great diversity in the literature regarding the approach to the topic. Nevertheless, there are descriptions of deterioration of the secondary and tertiary curves of the spine with growth in cases of congenital kyphosis. Over time, this can cause recurrent skin lesions, located at the apex of the kyphosis, as well as difficulty in balance and sitting ability. There may also be collapse of the lower portion of the rib cage onto the thighs, leading to respiratory compromise[8].

CONCLUSION

Congenital kyphosis is a condition rarely described in the literature in fetuses during the prenatal period. Additionally, it is noted that there is almost no description of cases evaluated, not only by fetal ultrasound but also by fetal MRI. As we have seen, the latter examination can serve as a complement to the former, for a better understanding, both of congenital kyphosis and also of related anomalies, such as the meningomyelocele observed in our fetus. In the present report, MRI also assisted in the planning of the delivery and the surgery to be performed immediately afterward. Thus, the importance of a combined approach, using both examinations, is emphasized, with the aim not only of diagnosing and improving the understanding of the alteration presented by the patient but also of identifying possible associated abnormalities and offering the best conditions for management and treatment.

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REFERENCES

1. Mintz LJ, Sarwark JF, Dias LS, Schafer MF. The natural history of congenital kyphosis in myelomeningocele. A review of 51 children. Spine (Phila Pa 1976). 1991;16(8):S348-350.
2. Zeng Y, Chen Z, Qi Q, Guo Z, Li W, Sun C, Liu N. The posterior surgical correction of congenital kyphosis and kyphoscoliosis: 23 cases with minimum 2 years follow-up. Eur Spine J. 2013;22(2):372-378.
3. Egloff A, Bulas D. Magnetic resonance imaging evaluation of fetal neural tube defects. Semin Ultrasound CT MR. 2015;36(6):487-500.
4. Beals RK, Robbins JR, Rolfe B. Anomalies associated with vertebral malformations. Spine (Phila Pa 1976). 1993;18(10):1329-1332.
5. Broekman BA, Dorr JP. Congenital kyphosis due to absence of two lumbar vertebral bodies. J Clin Ultrasound. 1991;19(5):303-305.
6. Song TB, Kim YH, Oh ST, Byun JS, Kim EK. Prenatal ultrasonographic diagnosis of congenital kyphosis due to anterior segmentation failure. Asia Oceania J Obstet Gynaecol. 1994;20(1):31-33.
7. Coleman BG, Langer JE, Horii SC. The diagnostic features of spina bifida: the role of ultrasound. Fetal Diagn Ther. 2015;37(3):179-196.
8. Marks DS, Qaimkhani SA. The natural history of congenital scoliosis and kyphosis. Spine. 2009;34(17):1751-1755.
9. Bakaloudis G, Lolli F, Di Silvestre M, Greggi T, Astolfi S, Martikos K, Vommaro F, Barbanti-Brodano G, Cioni A, Giacomini S. Thoracic pedicle subtraction osteotomy in the treatment of severe pediatric deformities. Eur Spine J. 2011;20(Suppl 1):S95-104.
10. Kawahara N, Tomita K, Baba H, Kobayashi T, Fujita T, Murakami H. Closing-opening wedge osteotomy to correct angular kyphotic deformity by a single posterior approach. Spine (Phila Pa 1976). 2001;26(4):391-402.

ANDRIELLE SOARES
http://lattes.cnpq.br/5160573045349381
https://orcid.org/0000-0003-2428-7985

CAROLINA MAROLLI
http://lattes.cnpq.br/0282109000236500
https://orcid.org/0000-0001-6824-5213

LEONARDO RUBIN
http://lattes.cnpq.br/2376584264050307
https://orcid.org/0009-0002-7367-5984

VALENTINA BRACHT
http://lattes.cnpq.br/0650802744344193
https://orcid.org/0000-0003-0317-9117

MARCELLE DRAGHETTI
http://lattes.cnpq.br/9593679055296836
https://orcid.org/0000-0003-2380-8160

JORGE TELLES
http://lattes.cnpq.br/8253976877321806
https://orcid.org/0000-0002-8108-9638

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