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Review article

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Congenital lordosis and lordoscoliosis: state of the problem (literature review)

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Abstract

Introduction Congenital lordosis is a severe deformity of the spine in the sagittal plane, which develops due to malformation and segmentation of the dorsal part of the vertebrae and normal growth of the ventral part. The most common nosological variant of the defect is lordoscoliotic deformity of the thoracic spine, which often occurs due to syndromic defects (spondylocostal dysostosis) and systemic diseases of the musculoskeletal system (arthrogryposis). Along with the vertebral syndrome, the pathological complex in congenital lordosis of the thoracic or thoracolumbar spine includes thoracic insufficiency syndrome, which in the overwhelming cases leads to the development of respiratory failure caused by bronchial obstruction. The **purpose** of the study is to summarize information and analyze literature data on a rare pathology of the spine, which is clinically manifested by the formation of thoracic lordosis due to congenital anomaly in the development of the vertebrae. **Materials and methods** An analysis of the literature on the diagnosis and treatment of congenital lordosis and lordoscoliosis in children was carried out. The depth of the search was more than 100 years. Inclusion criteria: presence of lordotic deformity of the thoracic, cervicothoracic or thoracolumbar spine. **Results and discussion** Respiratory failure in this group of patients develops due to reduction in the anteroposterior size of the chest (an increase in the chest penetration index), a decrease in the height of the thoracic spine, changes in the mechanics of rib movements, bronchial obstruction, worsening ventilation and perfusion on the convex side of the deformity. The leading component of disability and impairment of the quality of life of patients is respiratory failure syndrome. A decrease in lung function, primarily due to reduction in VC, is directly proportional to the magnitude of the deformation of the spine and chest. Surgical correction of spinal and chest deformity is the main component of treatment for this group of patients. Surgical treatment consists in the mandatory multilevel mobilization of all three columns of the spine (preferably combined access) and bilateral resection of the ribs to reduce chest rigidity. Postoperative respiratory support is critical in respiratory failure. **Conclusion** Congenital spinal deformities in the form of thoracic lordosis and lordoscoliosis are extremely rare. The key aspects of treatment are the fight against respiratory failure and the selective choice of surgical technologies for deformity correction with a trend towards simultaneity of interventions. The literature review confirmed the insufficient knowledge on the issues of this pathology.

Keywords: congenital lordosis, lordoscoliosis, children, congenital deformity of the spine

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INTRODUCTION

Congenital lordosis is a severe deformity of the spine in the sagittal plane, which develops due to disorders of formation and segmentation of the dorsal part of the vertebrae but with normal growth of the ventral part [1, 2]. The most common nosological variant of the defect is lordoscoliotic deformity of the thoracic spine, which often occurs in syndromic conditions (multiple pterygium syndrome or Escobar syndrome) and systemic diseases of the musculoskeletal system (arthrogryposis) [3, 4, 5].

Along with the vertebral syndrome, the pathological complex in congenital lordosis of the thoracic or thoracolumbar spine includes thoracic insufficiency syndrome, which in most cases leads to the development of respiratory failure caused by bronchial obstruction [6, 7].

The key component in the management of this group of patients is early surgical correction of the deformity, which provides both the restoration of the sagittal and frontal balance of the body and the reduction of the Spinal Penetration Index [8].

Due to a relative rarity of malformations with lordosis of the thoracic spine, its studies are single and the publications mainly describe clinical cases or small clinical series. Works of domestic authors are completely absent. Therefore, we set the goal to summarize information and analyze literature data on a rare pathology of the spine, clinically manifested by the formation of thoracic lordosis caused by a congenital anomaly in the development of the vertebrae.

MATERIAL AND METHODS

We searched available literature sources including PubMed, GoogleScholar, eLibrary databases with the following keywords: “congenital lordosis”, “congenital lordoscoliosis”. The depth of the search was maximum and amounted to more than 100 years (1916–2020). The search resulted in 29 studies found.

RESULTS AND DISCUSSION

Currently, there are many theories of the origin of congenital deformities of the spine. They are based on disorders in the process of laying and formation of vertebral bodies at the stage of somatogenesis, which occurs at 4-8 weeks of embryonic development [10, 11]. The pathogenesis of congenital lordosis is a deformity that develops by normal growth of the ventral part of the vertebrae and disturbance in the dorsal segmentation. It may be accompanied by disturbance of segmentation at the level of the ribs. Abnormal segmentation of the vertebrae can be associated with more than 150 genetic disorders, syndromes, or developmental sequences [12]. We will analyze the genetic syndromes associated with the formation of thoracic lordosis in this article.

Genetic syndromes

In 1938, Jarcho and Levin reported cases of chest insufficiency due to anomalies of the vertebrae and ribs [4]. In 1966, Lavy et al. described a similar syndrome in a Puerto Rican family [13]; in 1969 Monseley and Bonforte [14] also described two cases in children who died under the age of one year from pneumonia. In 1991, Karsen et al. clarified the Jarcho-Levin syndrome (a disease that manifests itself as malformations of the vertebral bodies and associated ribs) and reported 61 cases (57 from literature sources and 4 their own), dividing it into two main subtypes (spondylocostal dysostosis and spondylothoracic dysostosis) with different survival rates associated with malformations and types of inheritance [15]. Patients with spondylothoracic dysostosis have deformities of the vertebral bodies and ribs that expand fan-shaped, but do not have significant deformities. It is an autosomal recessive trait and these patients have a higher mortality rate and a higher incidence of spinal cord defects. Patients with spondylocostal dysostosis have spinal malformations, frequent rib malformations, and short stature, but do not have a fan-shaped chest. In most cases, spondylocostal dysostosis is inherited in an autosomal recessive manner, but in some families it is a dominant trait that correlates with better survival. The absence of early respiratory support is typical, which was the cause of high mortality in this group of patients, but the improvement of technologies for supporting the respiratory function of patients has increased survival.

Berdon et al. distinguish between these two syndromes in 2011 based on the type of inheritance and clinical and radiological picture [16]. Spondylocostal dysostosis (SCD) or Jarcho-Levin syndrome causes mild to moderate respiratory failure, is pan-ethnic, and is associated with a gene such as DLL3, which is known to be associated with the Notch pathway. Spondylothoracic dysostosis (STD) or Lavy-Moseley syndrome results in more severe respiratory disorders, is strongly associated with Puerto Rican cohorts, and is believed to be related to the MESP2 gene, also a Notch pathway gene. One of the manifestations of spondylocostal and spondylothoracic dysostosis is thoracic hyperlordosis, but these are rare genetic diseases.

History of understanding and treatment of thoracic lordosis

The first case of congenital hyperlordosis of the thoracolumbar location was described in 1916 by Kleinberg S. in a healthy child [9]. The deformity developed without concomitant anomalies and was not accompanied by a neurological deficit. Further information about the patient is not known.

The pioneer in the treatment of thoracic lordosis is Winter R.B., who in 1975 stated a negative effect of thoracic lordosis on lung function in idiopathic scoliosis compared to kyphoscoliosis [17]. And already in 1978, Winter et al. presented the results of surgical treatment of five patients with lordosis of the thoracic spine in combination with a reduced VC [18]. Four corrective surgeries were performed using anterior release and bracing, and one patient underwent dorsal fusion followed by bracing. Instrumental fixation was not used. Two patients died within 2 years after the operation. One patient developed tetraplegia. Two patients survived. In no case did the surgical treatment lead to the formation of thoracic lordosis. An additional problem is that during lordosis of the cervical region, problems with swallowing solid food may occur, i.e. dysphagia develops. The use of Halo traction leads to the formation of proximal adjacent kyphosis at the level of the cervicothoracic junction with the formation of a neurological deficit in the form of tetraplegia, which excludes the possibility of using this treatment option. Based on the presented data, the authors come to the following conclusions: pulmonary function is impaired in the absence of vertical growth of the spine and penetration of the spine into the chest from the front. They also stated that the earliest possible correction of the deformity is required, because treatment of patients aged 22 and 18 years ended in death.

Bradford D.S. et al. (1983) based on the treatment of three adult patients aged 17 to 24 years with severe thoracic lordoscoliosis described a combined surgical technique for the correction of thoracic lordosis with the restoration of normal kyphosis. The technique includes discectomy from the ventral approach, resection of the ribs for 10 cm (resection volume was 0.5-0.75 cm from the costovertebral angle) and dorsal sublaminar wire fixation to the Harrington rods in two stages. This technique in a small number of patients enabled to correct thoracic lordosis from 32º to the formation of thoracic kyphosis of 32º and increase the retrosternal distance

from 5 to 7.5 cm. This method provided an increase in chest volume and improved lung function [19].

In 1990 Winter R.B. et al. modified the operation technique proposed by Bradford D.S. [20] for the treatment of congenital pathology of the spine. The authors performed an anterior transthoracic wedge-shaped osteotomy (closed-wedge), additionally, in contrast to the previous technique; posterior osteotomy of laminar synostosis with deformity correction using kyphotic rods according to Luke E., followed by the procedure of "stretching" the lateral wall of the chest. This modification allows correcting thoracic lordosis in a total posterior bone block from 25º to the formation of kyphosis of 25º, as well as correcting scoliosis from 71 to 35º, which ensures an increase in VC from 740 to 1190 ml in the long term. In addition, the authors note that after the primary correction, the patient required forced ventilation for 6 days. Based on the foregoing, the authors concluded that congenital lordosis is a potentially lethal anomaly against the background of a vast area of laminar synostosis posteriorly and a large number of levels of growth in the front, which occurs due to progressive respiratory failure.

In 1992, Winter et al. reported another case of using this surgical technique, but for correction of neuromuscular lordoscoliosis [21]. They performed the correction of thoracic lordosis from 35º to the formation of thoracic kyphosis of 24º. In the postoperative period, positive ventilation was required for 8 weeks; the tracheostomy was removed after 10 weeks. Also in the postoperative period, intravenous nutrition and nutrition through a nasogastric tube were necessary. The patient switched to a normal diet 16 weeks after the intervention. All of the above indicates a high risk of postoperative respiratory complications and the severity of the surgical intervention, which requires high-quality anesthetic support in the peri-operative period.

In 1999, Lonstein summarized the available information on congenital spinal deformities and wrote a literature review based on 38 literature sources [22]. Based on the data, the author described the key features of the development and treatment of congenital lordosis:

1) With an increase in lordosis, there is a decrease in the distance between the spine and the sternum and a change in the mechanics of the ribs during breathing. This leads to respiratory limitation, respiratory failure, and even premature death.

2) No conservative treatment for congenital lordosis plays a role, since the natural course of the disease progresses. Treatment in all cases is surgical.

3) The deforming force is the growth of the anterior column of the spine. Therefore, in all cases, an anterior approach is required;

4) The only method for correcting congenital lordosis is osteotomy of an unsegmented bar.

Thus, in each case requiring correction, a combined anterior and posterior approach is necessary. As far as these patients have restrictive lung disease, the anterior approach is more risky. If an element of pulmonary insufficiency is already present, these risks increase. In pulmonary artery overstress, surgery is probably contraindicated due to the high mortality in such cases.

In the early stages, only anterior fusion can be used. In the thoracic spine, it is necessary to perform bilateral resection of the ribs so that they do not interfere with the correction of the spine.

Due to respiratory dysfunction and bilateral rib resections, postoperative respiratory support is critical.

It is worth paying attention to an extremely rare variant of lordoscoliotic deformity of the thoracic spine in a postpneumoectomy syndrome in a 17-year-old child, which was described by Godsi M.J. et al. (2000) [23]. Due to congenital pulmonary artery agenesis and recurrent respiratory distress syndrome, the girl underwent a right-sided pneumoectomy at the age of 14. Three years later, she was diagnosed with a shift of the mediastinal organs to the right, stenosis of the main bronchus of the only left lung, reduced VC and FVC parameters, left-sided heart failure, a progressive decrease in the vertebral-sternal space (later described by Dubousset J. as "chest penetration index") and a low magnitude of physiological thoracic kyphosis. To correct the defects, staged surgical treatment was performed with sealing of the mediastinal pleura with a mesh implant, polysegmental wedge-osteotomy of the anterior column, and instrumental fixation of the spine with a laminar structure in order to create thoracic kyphosis.

In 2001, Gogus A. et al. presented two clinical cases of surgical treatment of congenital thoracic lordosis and concluded that bilateral rib resection is mandatory using the combined technique of Bradford D.S. (1983) and Winter R.B. (1990) [24]. Correction of the thoracic lordosis was 79.5º, which led to the formation of kyphosis at an average of 18.5º. The authors state the need for surgical treatment in one surgical session, because two-stage surgery requires long-term postoperative respiratory support.

Dodson C.C. et al. (2005) reported the result of surgical treatment of a 17-year-old girl with multiple pterygium syndrome or Escobar syndrome which may also occur with the formation of lordoscoliosis. To correct the balance of the body, the patient underwent a one-stage polysegmental osteotomy with resection of the ribs and posterior instrumental fixation of the spine at the C7-L3 level [25]. The functional vital capacity of the lungs increased from 23 to 60 %. This may indicate that patients with severe spinal deformity secondary to Escobar syndrome respond successfully to surgical treatment. Preoperative planning by a multidisciplinary

team and meticulous surgical technique are required to ensure balanced deformity correction and restoration of lung function.

Dubousset J. et al. published the largest clinical series in 2003, represented by 16 cases of lordoscoliosis of the thoracic spine, combined with chest defects [8]. Based on 3D modeling and deformity analysis, the authors propose g the Spinal Penetration Index to assess the severity of both the vertebral and thoracic components of the deformity. This led to the division of deformities of the chest and spine into 3 groups: with the formation of an endothoracic vertebral hump, an exothoracic costal hump, and its analogue, an exothoracic "absent hump". Based on this classification, the tactics and scope of surgical treatment in each group were formed.

Thereby, the analysis of the pulmonary dysfunction severity due to congenital lordosis and lordoscoliosis, presented by Bartlet W. et al. (2009) reveals a direct relationship between this deformity and the development of focal bronchial obstruction, the severity of which progressively decreases after surgical correction of the spinal deformity [26].

In 2016 Kanagaraju et al. presented a case of treatment of congenital lordoscoliosis complicated by type 1 SCM. The treatment was carried out in 4 stages: Halo traction, Halo femoral traction for 4 weeks, then Halo pelvic traction and PSO at 2 levels, after 4 weeks final transpedicular fixation with correction of thoracic lordosis to 0º [27].

In 2017 Ha et al. published a clinical case of treating a patient with congenital lordoscoliosis [28] using PMVO osteotomy (posterior multilevel vertebral osteotomy, Schwab 2 according to the generally accepted classification of osteotomies), which allows the formation of thoracic kyphosis of 5º [29]. They also observed a continued growth of the spine after surgical treatment (Th1-L2 202 mm before surgery and 210 mm 2.5 years after surgery).

Also in 2017, a study by Sever et al. showed a series of clinical cases of 6 patients using a three-column osteotomy (PVCR) at several levels (from 1 to 3 levels) to correct congenital lordoscoliosis of the thoracic spine, which allows the formation of a thoracic kyphosis of 18.8º. They observed an increase in the anteroposterior size of the chest by 30 % [30].

In addition to the combined variants of the defect described above, the literature presents a case of the development of lordosis in arthrogryposis [5]. Imagama S. et al. (2013) used a combined approach, right-sided thoracotomy, polysegmental osteotomy of the spondylodized anterior column in combination with lobectomy and posterior instrumental fixation of the spine to correct the deformity associated with bronchial obstruction and atelectasis of the lower lobe of the right lung.

The formation of cervical hyperlordosis in Apert's syndrome [31] and congenital hyperlordosis of the cervicothoracic region lead to the formation of dysphagia, accompanied by the inability to close the epiglottis, which leads to bolus passage of food, compression of the posterior pharyngeal wall and a decrease in the elevation of the larynx, resulting in accumulation of food in the epiglottis and airway aspiration [32].

Given the severe course of congenital lordosis in the thoracic region, its rare occurrence and little knowledge of this pathology, further study of this pathology is required. It is necessary to develop unified approaches to surgical correction and a multicenter approach to pre- and postoperative management of patients in this nosological group, and consider the current trends in the correction of respiratory failure. It is necessary to generalize the accumulated experience in further work on this issue.

SUMMARY

The formation of congenital lordotic deformity of the thoracic region can be associated with both genetic pathology and develop due to isolated disturbance of segmentation of the posterior column of the spine. The deformity is inevitably accompanied by respiratory failure due to a decrease in the retrosternal distance, in the length of the thoracic spine, and a decrease in pulmonary artery perfusion. When the deformity spreads to the cervical spine, dysphagia may develop. Due to the rarity and lack of knowledge of congenital lordosis of the thoracic spine, the tactics of surgical correction were improved on the basis of data from the treatment of lordoscoliotic deformities of other etiology. A multidisciplinary approach is required to exclude and correct possible postoperative complications. Surgical correction requires mobilization of all three columns of the spine along the length and resection of the ribs to reduce chest rigidity. Treatment of patients in one surgical session prevents the development of severe respiratory failure. Based on the foregoing, the following conclusions can be drawn:

• Respiratory failure in this group of patients develops due to a decrease in the anteroposterior size of the chest (an increase in the chest penetration index), a decrease in the height of the thoracic spine, changes in the mechanics of rib movement, bronchial obstruction, deterioration of ventilation and perfusion on the convex side of the deformity;

• The leading component of disability and impairment of the quality of life of patients is the syndrome of respiratory failure;

• Decrease in lung function, primarily due to a reduction in VC, is directly proportional to the magnitude of the deformation of the spine and chest;

• Surgical treatment consists in the mandatory multilevel mobilization of all three columns of the spine (combined approaches are preferred) and bilateral resection of the ribs to reduce chest rigidity;

• Multi-stage interventions with different types of traction are not effective (deformation of adjacent levels with irreversible neurological symptoms may develop);

• Surgical correction of spinal and chest deformity is the main component of treatment for this group of patients;

• Postoperative respiratory support is critical in respiratory failure;

• Hyperlordosis at the level of the cervical spine can lead to dysphagia.

CONCLUSION

Congenital spinal deformities such as thoracic lordosis and lordoscoliosis are extremely rare. The key aspects of treatment are the fight against respiratory failure and the selective choice of surgical technologies for deformity correction and simultaneous interventions. The literature review confirmed the insufficient knowledge of this pathology.

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