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Unusual congenital vertebral body anomaly, with lumbalisation of the s1 vertebra and intervertebral disc degeneration. A case report

The vertebrae of the spine are formed during development by segmentation of the precursor spine tissue, in a process called somitogenesis. In this process, segments of tissue called somites are formed in pairs surrounding what will eventually become the spinal cord. The most common type of failure of formation anomaly is a hemivertebra. This is where a portion of the vertebra is missing resulting in a small, triangular shaped "half vertebra" or hemivertebra. Hemivertebrae can be classified based on their relationship to the adjacent spine (segmented, semisegmented, nonsegmented). When several vertebral segments fail to separate bilaterally, a block vertebra results producing fused vertebral bones. Unilateral unsegmented vertebral bars are caused by the failure of segmentation only on the left or right side of the spine. In the involved area of the spine there is absent or abnormal growth potential due to an area of missing bone (formation defect) or missing growth plates (segmentation defect). This results in an area of absent growth potential in the vertebral ring, and the growth in the remainder of the vertebral ring disrupts the normal alignment of the spine, producing different types of deformities. Failures of formation or segmentation may occur on either the right or left side of the body resulting in "pure" scoliosis, or in the anterior or posterior elements resulting in "pure" kyphosis or lordosis, respectively. Combined deformities are most common, producing scoliosis and sagittal plane deformity (3, 4).

The aim of the study is to present the case report of unusual vertebral body anomaly, with intervertebral disc degeneration, and transient vertebra in spiral CT examination with spatial imaging, and discuss the congenital spine anomalies.

MATERIAL AND METHODS

The study discusses the case of a 30-year-old man with congenital anomaly of the shape of L5 vertebral body, with transient S1 vertebra and intervertebral disc degeneration, complaining about transient back pains, and temporarily filings of blocking, restriction of the movements in the lumbar area. CT spiral examination of the lumbo-sacral spine was performed with 2 mm thick collimation, pitch – 1. The MPR reconstructions, and spatial 3D images SSD were performed to present the complex morphology of the anomaly.

RESULTS

On axial CT images nucleus pulposus herniated dorsally at the level L4-L5 was visible (Fig. 1). The degeneration of the intervertebral disc at the level L5-S1 was seen (phenomenon vacuum) (Fig. 2). The MPR reconstruction reveals the presence of the anomaly of the superior surface of the S1 vertebra, with bone process on it, the same bone process on the inferior surface of the L5 vertebra, with hole on the corresponding L5 superior surface (Fig. 3). The SSD image reveal the

presence of the transient vertebra S1 (Fig. 4). The SSD and VRT cut off images, projected from the inside, presented the complex anatomy of the anomaly (Fig. 5 and 6).

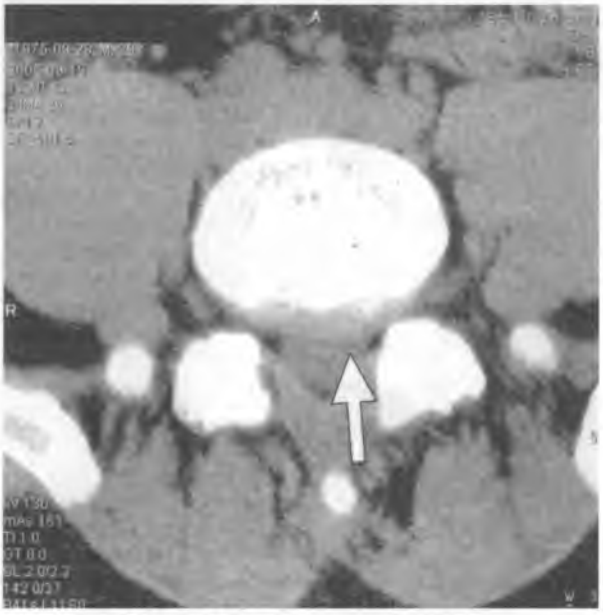

Fig. 1. Axial vertebral CT image revealing nucleus pulposus herniated dorsally (an arrow)

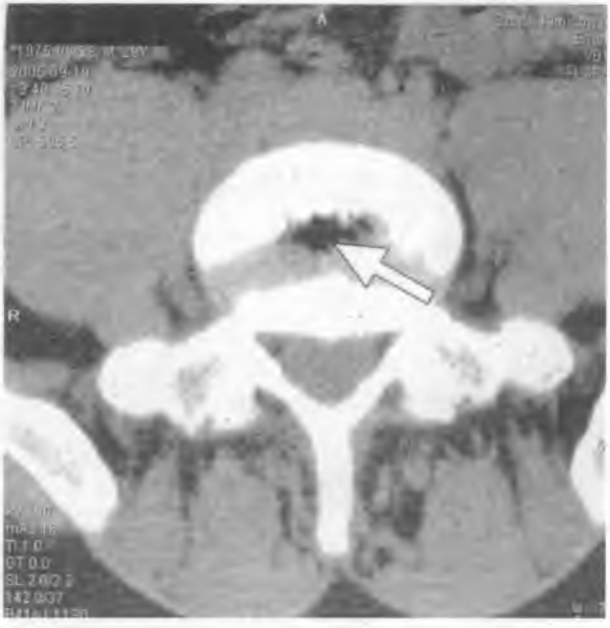

Fig. 2. Axial vertebral CT image revealing the degeneration of the intervertebral discus (phenomenon vacuum – an arrow)

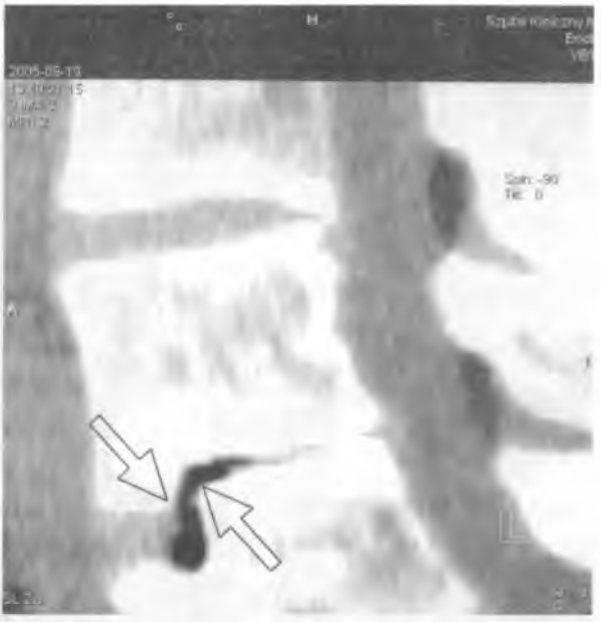

Fig. 3. MPR spine reconstruction vertebral bodies shape anomalies (arrows) resulting in the degeneration of the intervertebral discus

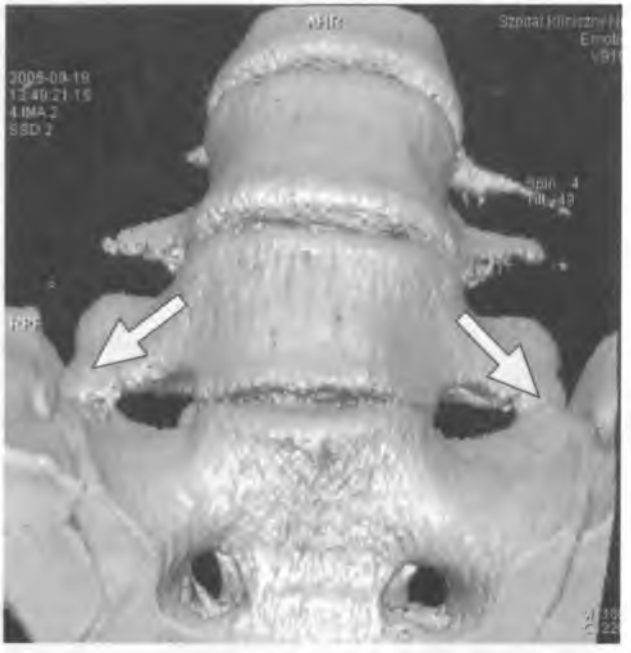

Fig. 4. SSD 3D image revealing the lumbalisation of the S1 vertebra

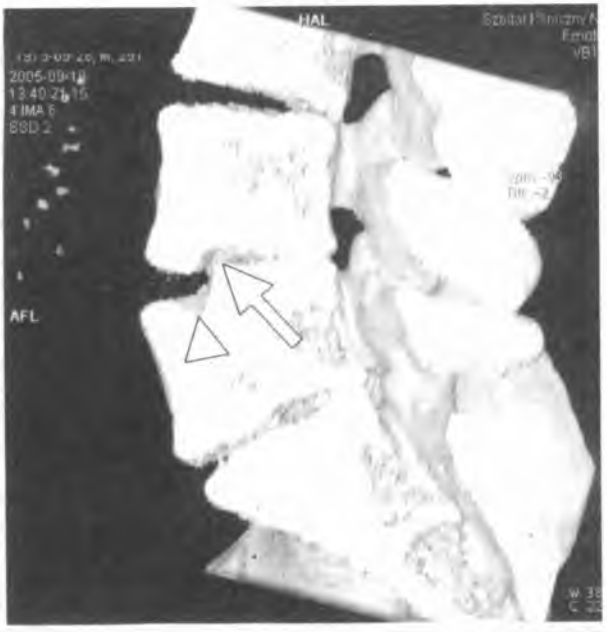

Fig. 5. SSD 3D image cut off along the spinous processes projected from inside reveal the S1 vertebral body anomaly, with additional bone structure on the superior surface (arrow) and the hole in front of it (arrow head)

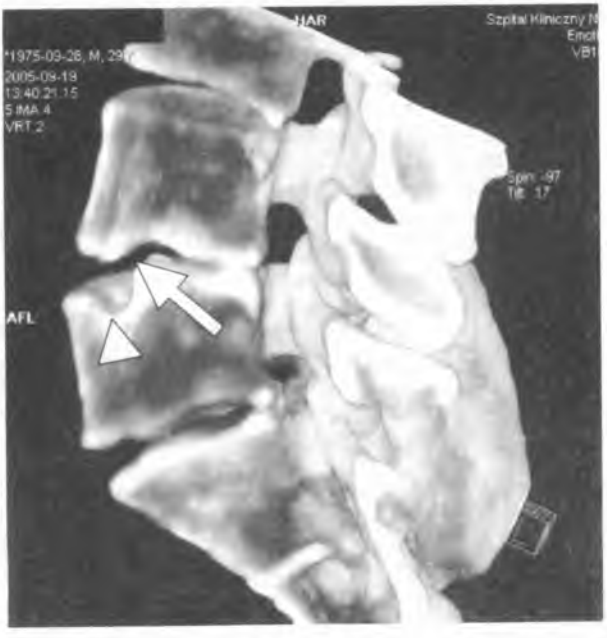

Fig. 6. VRT reconstruction of the previous image

DISCUSSION

Although congenital vertebral abnormalities are common, symmetric fusion defects leading to butterfly vertebra are not frequent in the general population. This defect is considered to occur between the third and sixth week of gestation. Developing vertebral bodies have two lateral chondrification centers that would normally fuse. If one fails to develop completely, a hemivertebra results. Failure of the two centers to fuse, however, results in the formation of a butterfly vertebra. A butterfly vertebra may associate with a few syndromes and chromosome deletions. It may be asymptomatic for long periods, but when discovered other body systems are carefully evaluated that may have been affected during embryogenesis.

A butterfly vertebra is said to be the result of a sagittal cleft caused by the persistence of the notochord. It is often associated with rib and/or vertebral anomalies such as bars, supernumerary lumbar vertebrae, and spina bifida. Butterfly vertebrae are seen in patients with Pfeiffer's syndrome, Jarcho-Levins (spondylothoracic or spondylocostal dysostosis) Syndrome, Crouzon Syndrome, Alagille Syndrome. Most cases with diastematomyelia are seen in association with other anomalies of the vertebral column such as spina bifida, kyphoscoliosis, and butterfly vertebra (6).

Spina bifida. Spina bifida is the most common neural tube defect, resulting from the failure of the fusion of the embryonic vertebral arches. The type of spina bifida is determined by the pattern of involvement of the vertebral arch, spinal cord, meninges and overlying dermis. The spinal cord defect may present at any vertebral level with the most common being the lumbosacral region, which is associated with the final component of neural tube closure. In addition to the structural abnormality, various other neurological defects may be present in the patient. These include hydrocephalus, diastematomyelia, Arnold-Chiari malformation, hydromyelia or a tethered spinal cord. Other signs of abnormalities may involve muscular balance, changes in bladder and bowel habits, sensory loss in the lower extremities and possibly paraplegia. In addition, many patients present with dislocation of the hips, clubfeet, scoliosis or kyphosis. A classification system has been established to better characterize the types and severity of the spina bifida.

Spina bifida occulta. Spina bifida occulta results from the failure of fusion or development of part of the vertebral arch, usually lamina, which does not involve the spinal cord or meninges. Several studies report an incidence of approximately 10% to 24% in the population. The studies state that this defect is usually asymptomatic, with only a small number of children showing concomitant defects in the spinal cord. Children with this defect usually present with a skin indentation and a patch of hair growing in the area of the lesion. However, because this defect may not present with an external manifestation, X-ray examination is the only valid test to confirm this type of neural tube defect. If the patient shows no associated abnormalities, no further treatment is necessary (5).

Spina bifida cystica. Spina bifida cystica is considered to be a severe defect, because it may involve the meninges as well as the spinal cord. Thus, several named subclasses of this defect exist. When the cyst involves the meninges and cerebrospinal fluid, it is referred to as spina bifida with meningocele. When the cyst also contains the spinal cord, the defect is referred to as spina bifida with meningomyelocele (5).

Spina bifida with meningocele. Patients developing spina bifida with meningocele usually have a layer of normal epidermis covering the meninges. This lesion does not involve the spinal cord, and therefore these patients usually do not present with any neurological symptoms. External manifestations that may indicate this type of neural tube defect include hair growth in the area of the lesion, lipomas, cysts or hemangiomas (5).

Spina bifida with meningomyelocele. Spina bifida with meningomyelocele is more common than spina bifida with meningocele. It is considered a more severe neural

tube defect because of the involvement of the spinal cord and meninges. This lesion may be covered by a thin layer of the skin or by a membranous sac. Patients with this lesion exhibit typical neurological symptoms, such as limb paralysis as well as bladder and bowel incontinence and may also present with hip dislocations. Because this defect may involve just the nerve roots or the entire spinal cord, paralysis may be of the flaccid, spastic or mixed type. Other associated abnormalities may include hydrocephalus, Arnold-Chiari malformations and severe forms of scoliosis, kyphosis or lordosis (5).

Myeloschisis. Myeloschisis is considered to be the most severe type of spina bifida. This defect occurs as a result of the neural tube not closing properly with the developed spinal cord being exposed to the external environment. Small patches of hair grow around the defect. As with spina bifida with meningomyelocele, involvement of the spinal cord translates into the possibility of one or more of the aforementioned associated defects, in addition to severe infection. Causes of neural tube defects are thought to be multifactorial in nature. According to several studies, genetic contributions appear to be negligible, but future studies may reveal the role of inheritance in neural tube defects. A slightly elevated risk of neural tube defects exists in siblings, but the data for risk regarding other relatives have yet to be proven. More important to the etiology of this defect is the environment (5).

Transitional vertebra. Transitional vertebrae are frequently encountered developmental variants of the spine. They are found in approximately 20% of human skeletons and often involve the sacrococcygeal and lumbosacral junctions. The L5 vertebra can be incorporated into the sacrum (i.e., "sacralized"), or the S1 vertebra can be incorporated into the lumbar spine (i.e. "lumbarized"). The transitional vertebra retains partial features of the segments above and below it so that the total number of vertebrae in the spinal column remains relatively constant. Transitional vertebrae are usually incidental findings during radiologic evaluation but rarely, when an enlarged transverse process irritates adjacent tissues, can be symptomatic. However, the most clinically important aspect of transitional vertebrae is the potential for confusion over the labelling or assignment of vertebral levels during medical or surgical treatment planning. Thus, to avoid inappropriate treatment of the wrong vertebral or disk space level, complete imaging evaluation of the spinal axis with fluoroscopy, radiography, or MR is required (2).

Congenital scoliosis. An abnormal vertebral development that results in a lateral curvature of the spine is classified as congenital scoliosis. This type of spinal deformity occurs through a failure of formation or a failure of segmentation. However, combined defects are the most common in congenital scoliosis. The type and region of the malformation determines severity of the scoliosis. In addition, a sagittal deformity can be involved, creating either a kyphoscoliosis or a lordoscoliosis. As with the other congenital spinal abnormalities, the defects appear to be sporadic, and studies have suggested a multifactorial basis, involving genetic and environmental contributions. As a result of the failure of cleavage of a primary center, part of the vertebra or growth plate will be unable to form and subsequent normal growth on the contralateral side will create the lateral curvature. In the case of a hemivertebra causing the scoliosis, an extra rib is a common additional finding. Eleven percent of the cases are seen in the lumbar region, with the remaining 5% in the lumbosacral region. Although unusual, congenital scoliosis does occur in the cervical or cervicothoracic regions as well (1, 5).

Congenital kyphosis. Congenital kyphosis refers to a deformity in the sagittal plane resulting in an excessive flexion of the affected area. Defects of this kind are classified as failures of formation, segmentation or dislocation of the spine as a result of rotation. As discussed previously, kyphosis with an associated scoliosis may be observed. The cause of this defect is also considered multifactorial in nature. In congenital kyphosis, a formational defect involves a complete or partial lack of a vertebral body. Deformities that are considered the most progressive usually have much more severe anterior defects. Congenital kyphosis may involve a partial failure

of formation of a vertebral element with a canal that is still properly aligned. This defect usually involves the posterior arch as well. Finally, there can be a complete failure of formation of a vertebral body. In this case, paraplegia is usually evident at birth and can be found for the most part in the lumbar spine. Defects of formation usually involve one level but can involve multiple levels. Most defects of formation occur in the thoracic or thoracolumbar segments of the spine and can encompass two to eight levels. As a result, the patient may present with a congenital spondylolisthesis resulting from instability of the spine. Most physicians agree that the most severe congenital kyphosis involves a failure of formation and a concomitant scoliotic curve. Failures of segmentation typically encompass more than one level and usually present as an unsegmented bar. The defect can be symmetrical or can show prevalence laterally. In the latter case, the patient presents with a kyphoscoliosis. The kyphosis may have a sharp or smooth angle depending on normal growth of the posterior elements and the extent of the congenital abnormality (2, 5).

Congenital lordosis. This defect is very uncommon and the least severe of the three abnormal curvatures discussed in this review. Defects of this kind create an abnormal extension of the spine. The only embryological defect associated with true congenital lordosis is a defect of segmentation posteriorly. However, there can be a simultaneous scoliosis creating a lordoscoliosis. Three factors may contribute to congenital lordosis. The first factor mentioned is the posterior defect of segmentation with concomitant normal anterior development. The second factor involves abnormal or lack of formation of the posterior elements. Spinal dysraphisms with defects in the posterior elements can be a primary cause of congenital lordosis. Finally, congenital lordosis may be a compensatory deformity as a result of a kyphosis at a lower vertebral level. As with other congenital vertebral defects, various systems may be involved. In the cardiopulmonary system, a severe lordosis can cause both constriction and stretching of the bronchi leading to atelectasis. These patients will present with severe pulmonary distress. In addition, typical abnormalities of the genitourinary and cardiac systems may be present in these patients. Patients with congenital lordosis also present with nervous system malformations. This condition is considered rare, but because of associated secondary abnormalities, it may develop into a severe defect when left untreated (5).

Klippel-Feil syndrome. Klippel-Feil syndrome or brevicollis involves the congenital fusion of two or more cervical vertebrae. This abnormality is the result of a failure of proper segmentation of vertebrae in the cervical region during embryonic development. In addition, theories suggest that defects in the notochord and notochord signalling may also cause this syndrome. Patients presenting with this defect have congenitally fused cervical vertebrae causing a shortened neck with a low posterior neckline and a diminished ability to move in the affected area. Patients are restricted to flexion and extension between the occiput and atlas. Klippel-Feil syndrome is classified into three categories: Type I, II and III. In Type I, the patient presents with numerous fused cervical vertebrae and possibly upper thoracic vertebrae with synostosis. Type II includes fusion of one or two vertebrae and other abnormalities of the cervical spine. Type III includes fusion of cervical vertebrae with concomitant fusion of thoracic or lumbar vertebrae. A fourth type addresses genetic heterogeneity and affected levels in the cervical spine. Patients with Klippel-Feil can present with an array of symptoms and may not present with the classic findings. However, it is important for the physician to recognize the syndrome in order to address the issue of cervical stability, any degenerative changes that may occur in lower cervical segments and other associated abnormalities (5, 7).

Congenital spondylolisthesis. Spondylolisthesis is a condition that involves a forward slip of a vertebra or vertebrae in relation to the rest of the spinal column. Typically, this defect occurs between the fifth lumbar vertebra and the sacrum. However, spondylolisthesis has also been seen between the fourth and fifth lumbar vertebral levels and may even occur in the cervical region. There are various types of spondylolisthesis. Thus, a classification system was established to differentiate the specific etiologies. The cause of congenital or dysplastic spondylo-

listhesis, which is most commonly found at the lumbosacral junction, occurs as a result of an elongation of the pars interarticularis along with varying degrees of facet joint dysplasia. Patients may present with back pain, radicular complaints, tight hamstrings and various neurological symptoms. The possibility of developing impairment of bowel and bladder function exists with high-grade neural compression. Neurological deficits necessitate prompt surgical intervention (5).

CONCLUSIONS

Development of the spine occurs through a complex series of events involving genes, signalling pathways and various metabolic processes. Because several systems develop from a common precursor, a defect early in fetal life may have a variety of clinical presentations. Physicians should be aware of these common types of congenital spinal defects as well as have an understanding of the normal embryologic process of development. Recognition of any of these defects indicates the need of a thorough physical examination to identify possible associated abnormalities in other systems as well. CT examination, especially with spatial imaging enables precise identification of the anomaly, and potential complication. The vertebral body shape is precisely and easily presented, especially in spatial SSD and VRT images provided information helpful in treatment planning.

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SUMMARY

The aim of the study is to present the case report of unusual vertebral body anomaly, with intervertebral disc degeneration, and transient vertebra in spiral CT examination with spatial imaging, and discuss the congenital spine anomalies. The study discusses the case of a 30-year-old man with congenital anomaly of the shape of L5 vertebral body, with transient S1 vertebra and intervertebral disc degeneration, complaining about transient back pains, and temporarily filings of blocking, restriction of the movements in the lumbar area. CT spiral examination of the lumbosacral spine was performed with 2 mm thick collimation, pitch – 1. The MPR reconstructions, and spatial 3D images SSD were performed to present the complex morphology of the anomaly. On axial CT images nucleus pulposus herniated dorsally at the level L4-L5 was visible. The degeneration of the intervertebral disc at the level L5-S1 was seen (phenomenon vacuum). The MPR reconstruction reveals the presence of the anomaly of the superior surface of the S1 vertebra, with bone process on it, the same bone process on the inferior surface of the L5 vertebra, with a hole on the corresponding L5 superior surface. The SSD image reveals the presence of the transient vertebra S1. The SSD and VRT cut off images, projected from the inside, presented the complex anatomy of the anomaly. Development of the spine occurs through a complex series of

events involving genes, signaling pathways and various metabolic processes. Because several systems develop from a common precursor, a defect early in fetal life may have a variety of clinical presentations. Physicians should be aware of these common types of congenital spinal defects as well as have an understanding of the normal embryologic process of development. Recognition of any of these defects indicates the need of a thorough physical examination to identify possible associated abnormalities in other systems as well. CT examination, especially with spatial imaging enables precise identification of the anomaly, and potential complication. The vertebral body shape is precisely and easily presented, especially in spatial SSD and VRT images provided information helpful in treatment planning.

Rzadka wrodzona anomalia kształtu trzonu kręgu z lumbalizacją kręgu S1 i degeneracją krążka międzykręgowego. Opis przypadku

Celem pracy jest opis przypadku rzadko spotykanej anomalii kształtu trzonu kręgu S1 u pacjenta z lumbalizacją S1, degeneracją krążka międzykręgowego, oraz przedstawienie typowych wrodzonych anomalii kręgosłupa. Praca omawia przypadek 30-letniego mężczyzny z rzadką wadą kształtu trzonu S1, jego lumbalizacją oraz degeneracją krążka międzykręgowego (*phenomen vacuum*). Badanie TK odcinka LS było wykonane tomografem spiralnym w odcinku LS, kolimacja skanów 2 mm, pitch 1. Oceniano skany osiowe, rekonstrukcje MPR oraz wtórnie wykonano rekonstrukcje przestrzenne SSD (*surface shadow display*) oraz VRT (*volume rendered technique*). Na przekrojach osiowych uwidoczniono centralną przepuklinę jądra miażdżystego krążka międzykręgowego L4-L5. Obrazy SSD przedstawiły krąg przejściowy S1 oraz zniekształcenie górnej blaszki granicznej z wyrośla kostną w części środkowej, wyrośla kostną w dolnej blaszce granicznej trzonu L5 i odpowiadające mu wgłębienie w przedniej części górnej blaszki granicznej S1. Zmiany kostne doprowadziły do degeneracji krążka międzykręgowego na poziomie L5-S1, z objawem próżniowym (*phenomen vacuum*). Obrazy SSD i VRT wyraźnie uwidoczniły opisywane anomalie kostne. Rozwój kręgosłupa jest skomplikowanym procesem, a jego zaburzenia na różnych etapach są przyczyną wrodzonych zmian morfologicznych. Często towarzyszą im zmiany wrodzone w innych narządach. Badanie TK umożliwia szczegółową ocenę wad i możliwych powikłań. Kształt kręgosłupa jest dokładnie i wyraźnie przedstawiony. Szczególnie przestrzenne rekonstrukcje SSD oraz VRT zapewniają informacje pomocne przy planowaniu dalszego postępowania leczniczo-rehabilitacyjnego.