SURGICAL TREATMENT OF PATIENTS WITH CAUDAL REGRESSION SYNDROME

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The paper presents clinical examples of surgical treatment of female patients aged 3 and 13 years having congenital malformations of the lumbosacral spine, sacral aplasia, and tethered spinal cord. The patients underwent surgical treatment, with due account for spine malformation and presence of tethered spinal cord syndrome.
Key Words: sacral aplasia, tethered spinal cord syndrome, treatment.

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According to the literature [1], the incidence of congenital malformations is 2 to 11 %. According to A.A. Gumerov et al. [4], the incidence of vertebral abnormalities accounts for not more than 3 % of all skeletal malformations. Some authors believe, that vertebral malformations of the lumbosacral spine belong to the most severe congenital pathology of the axial skeleton, since they can not be compensated in the lower segments and therefore lead to severe abnormalities of spinopelvic relationships [3]. Congenital malformation of the distal spine and spinal cord, the so-called caudal regression syndrome, is the most rare and severe abnormality [2]. According to S.I. Kozlova et al. [5], the population rate is 1 per 60 000 births, male to female ratio is 2.7:1. The disease is characterized by autosomal dominant inheritance. E.V. Ulrich et al. [7] studied the prevalence of spinal malformations in the population and noted that kyphogenic spine disorders are always dynamically unstable and the rate and extent of their progression directly correlate with the underdevelopment of the anterior and middle columns. In the case of vertebral agenesis, the incidence of neurological complications is more than 66.0 % (spinal canal stenosis is 2/3 or more of the proper values); in the case of hypogenesis it amounts to 33.0 %, and in the case of hypoplasia – 20.0 %.

The literature describes various ways to stabilize the spinopelvic segment in patients with caudal regression syndrome using metal structures with bone block formation [8–11]. In our opinion, Dunn-McCarthy lumbopelvic stabilization of the spine with metal structures and bone grafting using autogenous rib is the method of choice. This method is being promoted as an alternative option for fixation of sacrum and pelvis in children. Stable screw fixation is not always possible in patients with small size of iliac bones and sacrum. Specially shaped rods resting on the iliac wings in some cases may be an alternative to screw fixation. This fixation provides stability of the distal end of the metal structure, which is important to preserve the sagittal and frontal balance and establish strong fusion [6].

Well-developed tactics of preoperative examination and surgical treatment of patients with congenital malformations of the spine enables restoring global sagittal and frontal balance, maximum possible correction of deformity, restoring lumbar lordosis and spinopelvic relationships, whereby the child will develop harmoniously, and the quality of life will move to a new level.

Clinical case 1. Patient T., 13 years old, was examined and treated at the department of spine pathology of N.N. Priorov Central Institute of Traumatology and Orthopedics. She was diagnosed with "myelodysplasia, multiple lumbosacral malformations, type 3 caudal regression syndrome (according to Cama), congenital combined moderate scoliosis, dysfunction of pelvic organs". At admission, the patient complained of deformity and pain in the lumbosacral spine, rapid fatigability, and urinary incontinence.

Medical history. The child suffers from an isolated congenital malformation, chronic pyelonephritis, and bullous cystitis. The girl has maladaptive non-reflective bladder. It is known that spinal deformity and dysfunction of pelvic organs was detected at the age of 2 months. At the age of 9 months, she was diagnosed with *spina bifida* of the lumbosacral spine and myelodysplasia. The child was consulted at the Medical Genetics Research Center of the Russian Academy of Medical Sciences. There are no convincing data about hereditary skeletal abnormalities. Maximum progression of spinal deformity was observed at the age of 11 years. The child regularly received

conservative treatment at the place of residence in the form of massage, exercise therapy, and physiotherapy.

Dynamic observation of the patient revealed increase in forward inclination of body and formation of frontal and sagittal imbalance. The patient was admitted to the Department of Spine Pathology for further examination and development of surgical treatment tactics. At the department, the child underwent clinical and radiological examination.

Objective status. Conditions is relatively satisfactory. Natural color of skin and visible mucosa. Subcutaneous adipose tissue is not well defined. Palpable lymph nodes are not enlarged and painless. The rib cage is deformed. Percussion shows lung sounds over the entire surface of the lungs, auscultation shows puerile respiration with suppressed breath sounds and no rale. Respiration involves all parts of the lungs. Heart sounds are clear, rhythmical. The abdomen is involved in the breathing act; it is soft and painless during palpation. The liver is not enlarged, painless. The spleen is not palpable. The patient has dysfunction of pelvic organs (incontinence). Pediatrician's opinion: chronic pyelonephritis, bullous cystitis, non-adaptive non-reflective bladder.

Neurological status. Clear consciousness, active position, normal spatial orientation, the patient is cooperative. Pupils D = S, bilaterally positive photoreaction. Full-scale movement of the eyeballs. Palpebral fissures D = S, nasolabial folds are symmetrical. Swallowing and phonetics are not disturbed. Tongue at the midline. The patient satisfactorily performs coordination tests. Cranial nerves are intact, oral automatisms are absent, tendon reflexes D = S, hypotonia in the lower limbs, especially distal portions; no sensory disorders have been detected. Dysfunction of pelvic organs (urinary incontinence). Neurologist's opinion: myelodysplastic syndrome, pelvic dysfunction syndrome (incontinence), non-reflective bladder.

Orthopedic status: The patient can walk without assistance and additional support, with forward body inclination. Shoulder girdle, shoulder blades, and waist triangles are asymmetric. Posterior examination shows pronounced kyphosis of the lumbar spine. The axis of the spine is inclined to the right in its thoracic portion. Palpation along the spinous processes and paravertebral points is painless. Forward bending of the body is limited. Foot arches are flattened, there is valgus foot deformity (Fig. 1a). Orthopedist's opinion: multiple malformations of the lumbosacral spine, type 3 caudal regression syndrome (Cama). Congenital moderate combined scoliosis. Planovalgus foot deformity.

The results of X-ray diffraction (Fig. 2a) showed signs of combined dextroscoliosis, thoracic curve angle was 5°, lumbar – 12°. X-ray examination of the hip joints showed signs of dysplasia: femoral head is insufficiently covered with the acetabular roof.

There is positive sagittal balance up to 4 cm. According to CT scan (Fig. 2b), there is severe malformation of the lumbosacral spine due to abnormal junction of the vertebral bodies and arches along with bone defects of arch roots, hypoplasia of lumbar vertebral bodies, aplasia of sacral vertebral bodies (sacrum is represented by rudiments). The inferior lumbar vertebrae are moderately shifted towards the sacrum, posterior elements of the lumbar vertebrae are adjacent to each other to form separate bone blocks, spinal canal is expanded due to anterior dislocation of lumbar vertebral bodies and lordosis is enhanced. MRI results (Fig. 2c) showed signs of adhesions of the dura mater in the caudal spine and thickening of the terminal thread. Spirography shows that ventilation pulmonary function is not disturbed.

In view of the clinical and X-ray picture of the disease and recommendations of neurosurgeon, the patient first underwent transection of the thickened terminal thread at the L5–S1 level.

Skin was incised at the L5–S1 level under endotracheal anesthesia in the patient's prone position after triple treatment of the skin with antiseptic solutions. Posterior elements of L5–S1 vertebrae were skeletonized. Intralaminar space was extended due to partial resection of L5 and S1 pedicles. Revision of the epidural space revealed dorsally located thickened terminal thread. The dura mater was dissected linearly, incision length was 1 cm. Revision of the subdural spaces revealed dysplastic (lipomatous) stretched terminal thread. Meningoradiculolysis was performed using microsurgical techniques. The thread was treated using bipolar coagulation and 0.5 cm segment was excised; retraction of the thread in the cranial direction was noted. Hemostasis. We sutured the dura mater using encircling stitch and carried out tamponade with hemostatic sponge.

The postoperative period was uneventful. In 10 days, the second stage of the surgical treatment of spinal defor-

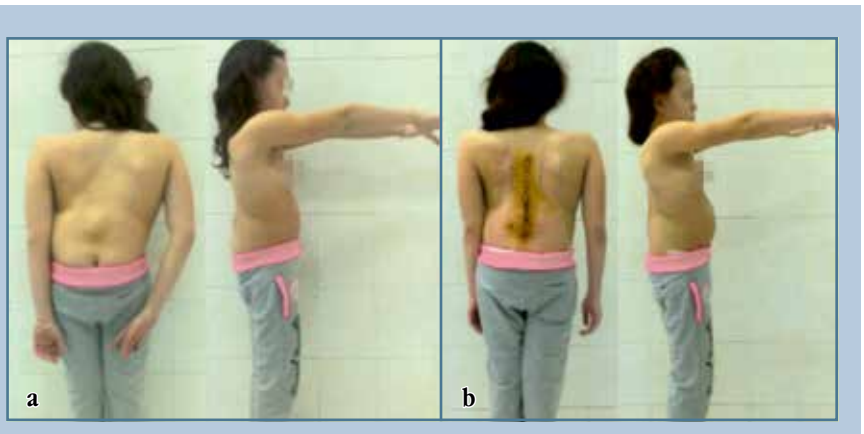

Fig. 1
Appearance of the patient T., 13 years old, before **(a)** and after **(б)** the operation

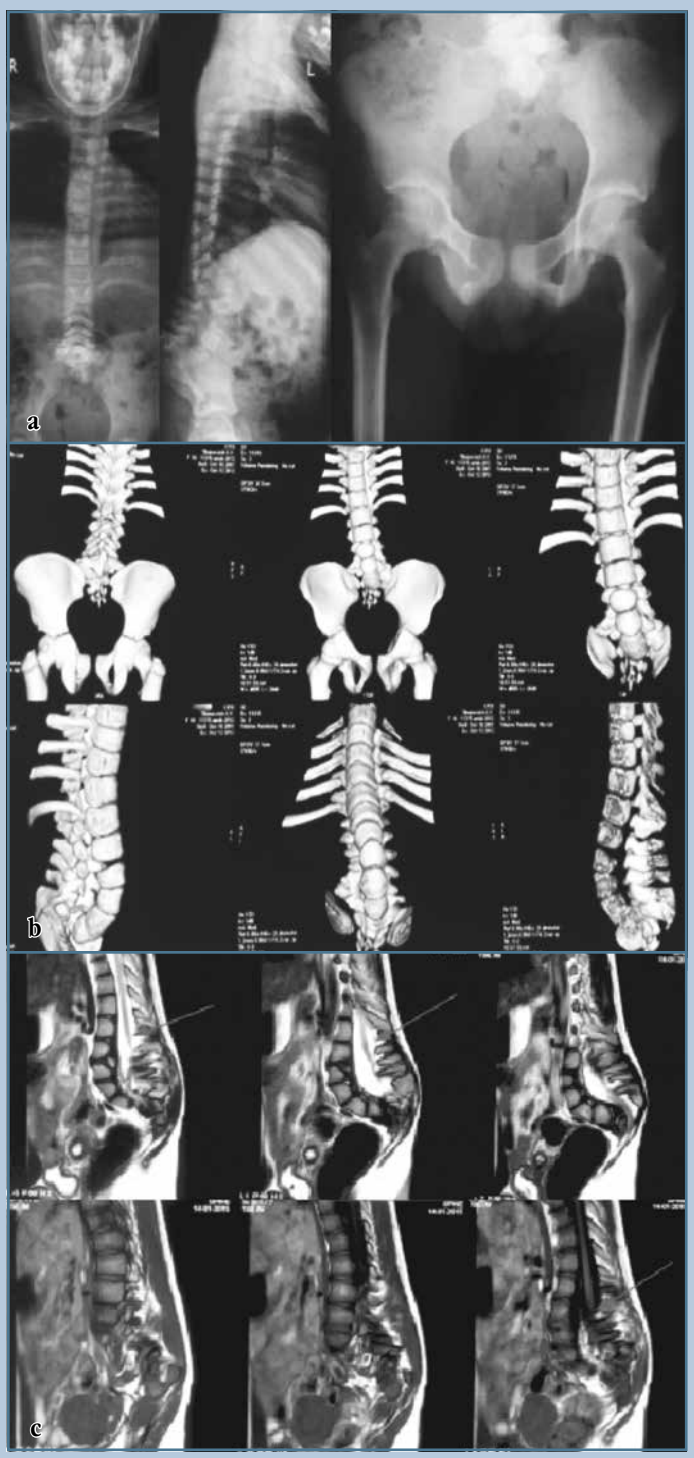

Fig. 2
X-Ray examination of the patient T., 13 years old, before the operation: **a** — frontal and lateral X-ray picture and X-ray picture of the hip joints; **b** – 3D CT; **c** – MRI (arrows indicate areas of lipodystrophy and thickened terminal thread)

mity was carried out, including dorsal correction and fixation of the thoracolumbar spine using metal structure under neurophysiological monitoring and bone grafting using allograft.

We placed electrodes to the target muscle, according to the operated spinal segment, under endotracheal anesthesia in the patient's prone position. After triple treatment of the skin with antiseptic solutions, we incised the skin at the level of T7-S1 vertebrae. Subcutaneous tissue and fascia were dissected layer-by-layer and posterior elements of the vertebrae were skeletonized on both sides. Hemostasis. Pedicle screws were mounted through the arc roots in the T8-T11 vertebral bodied on both sides. Two screws were mounted to the wings of the pelvis on each side. EOC-control: position of the screws is correct. Positioning of pedicle screws was assessed intraoperatively using neurophysiological monitoring. Rods designed with allowance for the deformity were laid on the screw heads on both sides. Segment-wise contraction and distraction was performed. Cross tie rods were mounted at the level of T12 and L2. Bone grafting with allografts was done (Fig. 3).

The postoperative period was uneventful. Antibacterial, anti-inflammatory, vascular therapy, and therapy with iron preparations was carried out. Patient was verticalized in the Leningrad-type thoracolumbar corset on day 3. The course of rehabilitation treatment in the form of physical therapy was carried out.

Postoperative X-ray study was performed (Fig. 4) and showed that the position of the metal structure is stable, the angle of the thoracic spine deformity is 1°, lumbar – 3°. Sutures were removed on day 14. After surgery, there was no worsening of neurological picture in lower extremities (Fig. 1b). The patient received recommendations and was discharged for outpatient monitoring.

The patient was questioned based on the standardized SRS-22 scale before the operation, 10 days and 6 months after operation. Before the surgery, the patient rated her appearance and the overall activity as 1.50 and 1.40 points, respectively. On day 10 after the operation, motor activity score was 2.25 points, satisfaction with the results of treatment was 4.20 points. After 6 months, this values increased: motor activity up to 2.55 points, satisfaction with the results of treatment up to 4.80 points, which is indicative of the increased quality of life of the patient.

Clinical case 2. Patient S., 3 years old, was examined and treated at the department of spine pathology of the N.N. Priorov Central Institute of Traumatology and Orthopedics. She was diagnosed with "malformations of the lumbosacral spine, type 1 caudal regression syndrome (according to Cama), and distal flaccid paraplegia. At admission, there were complaints of spine deformity.

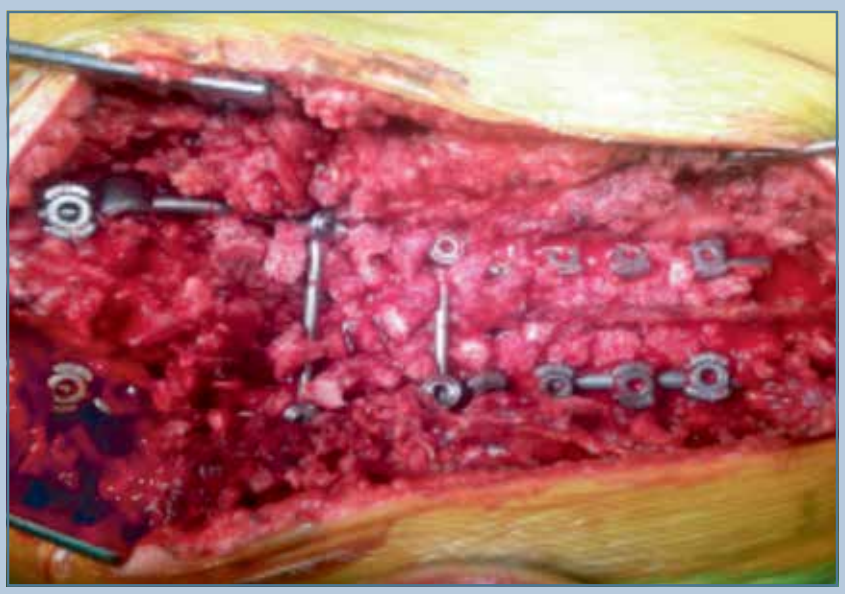

Fig. 3
The final stage of the operation in the patient T., 13 years old

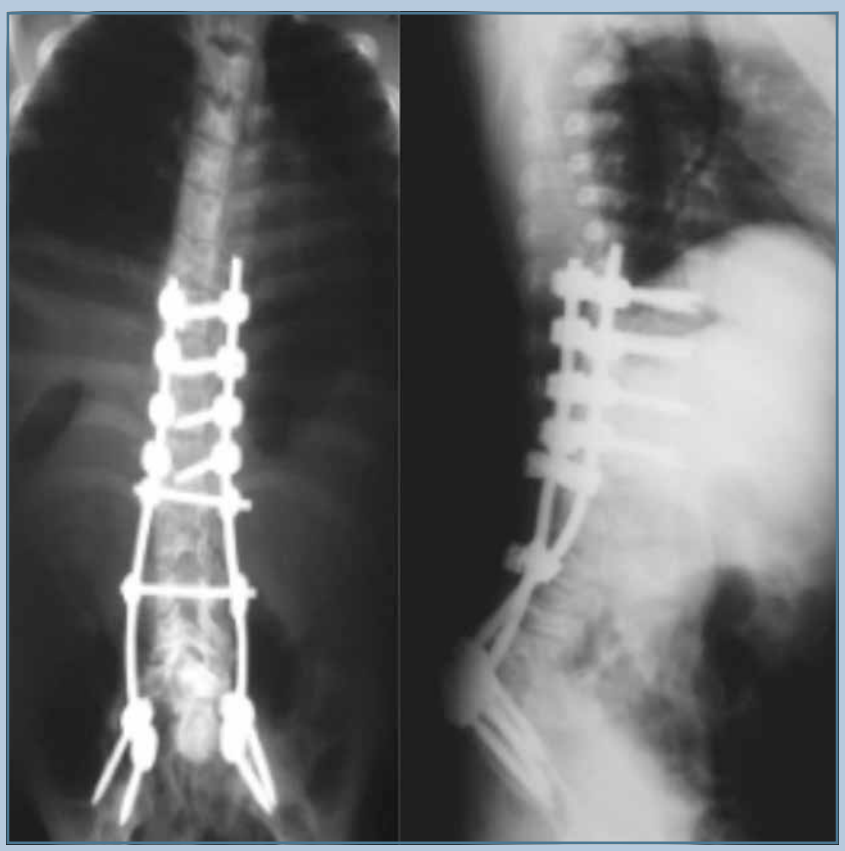

Fig. 4
X-ray pictures of the patient T., 13 years old, after surgery

Medical history. The child was followed up from birth by orthopedist at the place of residence for deformities and abnormal development of the lumbosacral spine and hypoplasia of the sacrum. Deformity progressed with growth of the child. The girl was admitted to the department of spine pathology for further examination and development of tactics of surgical treatment. At the department, the child underwent clinical and X-ray examination.

Objective status. Condition is satisfactory. Natural color of skin and visible mucosa. Subcutaneous adipose tissue is not well defined. Palpable lymph nodes are not enlarged and painless. The rib cage is of regular shape. Percussion shows lung sounds over the entire surface of the lungs, auscultation shows puerile respiration without rale, which involves all parts of the lungs. Heart sounds are clear, rhythmical. The abdomen is involved in the breathing act; it is soft and painless during palpation. The liver is not enlarged, painless. The spleen is not palpable. The patient has dysfunction of pelvic organs (incontinence). Pediatrician's opinion: dysfunction of pelvic organs (incontinence), chronic pyelonephritis.

Neurological status. Clear consciousness, active position, normal spatial orientation, the patient is cooperative. Pupils D = S, bilaterally positive photoreaction. Full-scale movement of the eyeballs. Palpebral fissures D = S, nasolabial folds are symmetrical. Swallowing and phonetics are not disturbed. Tongue at the midline. The patient satisfactorily performs coordination tests. Cranial nerves: pairs VII and XII are on the right, central type. Motor status: atrophy of tibial and gluteal muscles. Tendon reflexes: upper extremities – active and balanced, knee and Achilles reflexes can not be induced. Active movements in the feet and toes are missing. Plegia of gluteal muscles. Mixed dysfunction of pelvic organs: stool and urine retention, incontinence. Neurologist's opinion. Malformations of the lumbosacral spine, caudal syndrome, distal flaccid paraplegia.

Orthopedic status. The patient can walk with assistance of adults with her

body inclined forward, waddling-type walking. The chest is expanded. The pelvis is narrow. Shoulder girdle, shoulder blades, and waist triangles are moderately asymmetric. Posterior examination shows pronounced kyphosis of the lumbosacral spine. The axis of the spine is normal. Palpation along the spinous processes and paravertebral points is painless. Forward bending of the body is limited. Contractures of the ankle and knee joints are detected. Active movements in the feet and toes are missing. Foot arches are flattened, valgus foot deformity is observed (Fig. 5). Orthopedist's opinion: malformation of the lumbosacral spine, type 1 caudal regression syndrome (Cama), planovalgus foot deformity, contractures of the ankle and knee joints.

The patient was not questioned on a standardized SRS-22 scale due to young age.

The results of the x-ray examination (Fig. 6) showed signs of abnormal development of the lumbosacral spine: L5 vertebral body is significantly reduced in size, which is accompanied by abnormal junction. L5 vertebral body articulates with the posterior portions of the iliac wings, the volume of pelvis is reduced due to aplasia of the sacrum.

Taking into account clinical and x-ray presentation of the disease, patient underwent dorsal lumbopelvic stabilization of the spine using steel structure according to Dunn-McCarthy technique under neurophysiological monitoring and bone grafting using autologous rib.

We placed electrodes to the target muscles according to the operated spinal segment under endotracheal anesthesia in patient's prone position.

Skin was incised at the level of L3–L5 vertebrae along the line of spinous processes after triple treatment of the skin with antiseptic solutions.

Subcutaneous tissue and fascia were dissected layer-by-layer and posterior vertebral elements were skeletonized on both sides. Approach to the iliac wings was performed. Hemostasis. Pedicle screws were mounted through the arch roots to L4–L5 vertebral bodied. Beds for the rods were formed on both sides of the iliac wings. EOC-control: position of screws is correct. Positioning of pedicle screws was intra-operatively assessed using neurophysiological monitoring. Two rods designed with due account for lumbar lordosis were placed to the prepared bed on both sides and rested on the iliac wings. After triple treatment of the skin in the projection of the ribs 6 to 8, skin was incised above the seventh intercostal space on the left from the posterior axillary line to the inferior angle of the scapula. Subcutaneous tissue, fascia, and muscles were dissected layer-by-layer. Dorsal portions of the ribs 6 to 8 were isolated and subjected to subperiosteal resection 8 cm in length. The ribs were prepared, placed as autografts along the metal structure, and fixed with threads (Fig. 7).

The postoperative period was uneventful. We conducted antibacterial, anti-inflammatory, vascular therapy, and therapy with iron preparations. Patient was verticalized in Leningrad-type lumbosacral corset on day 3. Sutures were removed on day 14. No worsening of neurological picture in lower limbs was observed after surgery. The patient received recommendations and was discharged for outpatient monitoring.

Follow-up period for this patient was 2 years. Clinical examination revealed no loss of correction (Fig. 1b). X-ray control showed that metal structure is stable (Fig. 8). Neurological examination showed positive dynamics in the form of active movements in toes, regression of plegia of the gluteal muscles, and functional recovery of pelvic organs.

Conclusion

The use of modern stabilizing systems in combination with bone grafting enables correction and stabilization of the lumbosacral spine. This type of operation can improve the quality of life of patients with caudal regression syndrome.

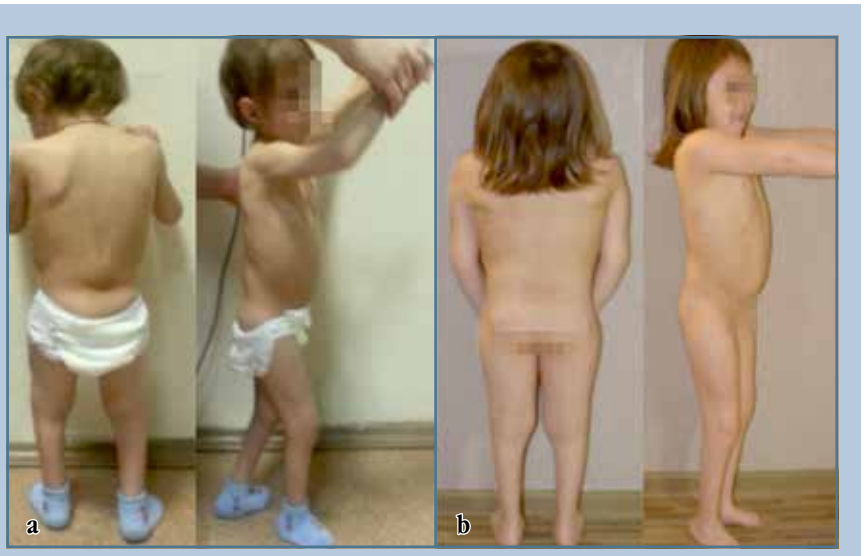

Fig. 5
Appearance of patient S., 3 years old, before **(a)** and 2 years after surgery **(b)**

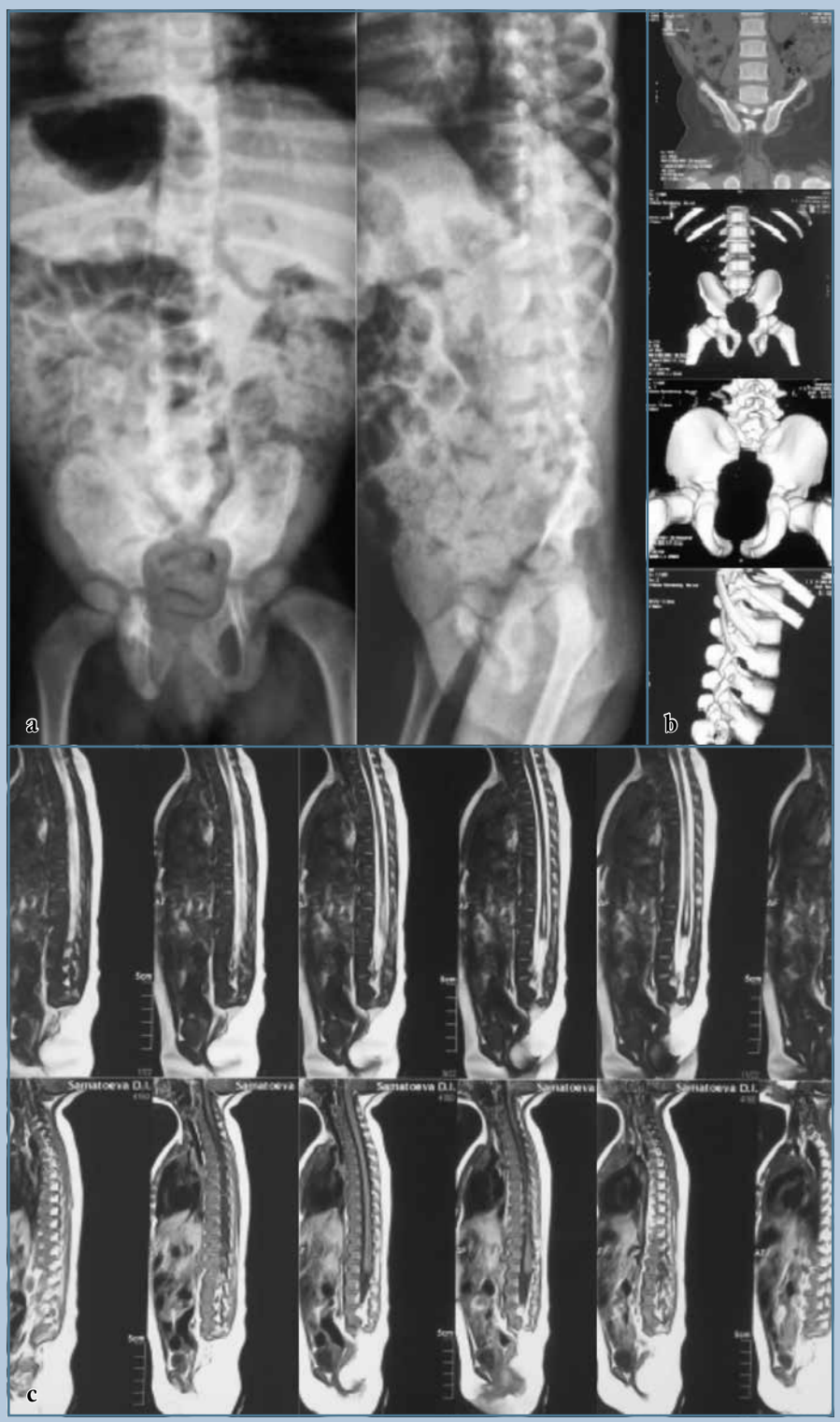

Fig. 6
X-Ray examination of the patient S., 3 years old, before the operation: **a** – frontal and lateral X-ray picture; **b** – 3D CT of the lumbosacral spine; **c** – MRI

Fig. 7
Stages of the operation in the patient S., 3 years old (autologous rub is shown by arrow)

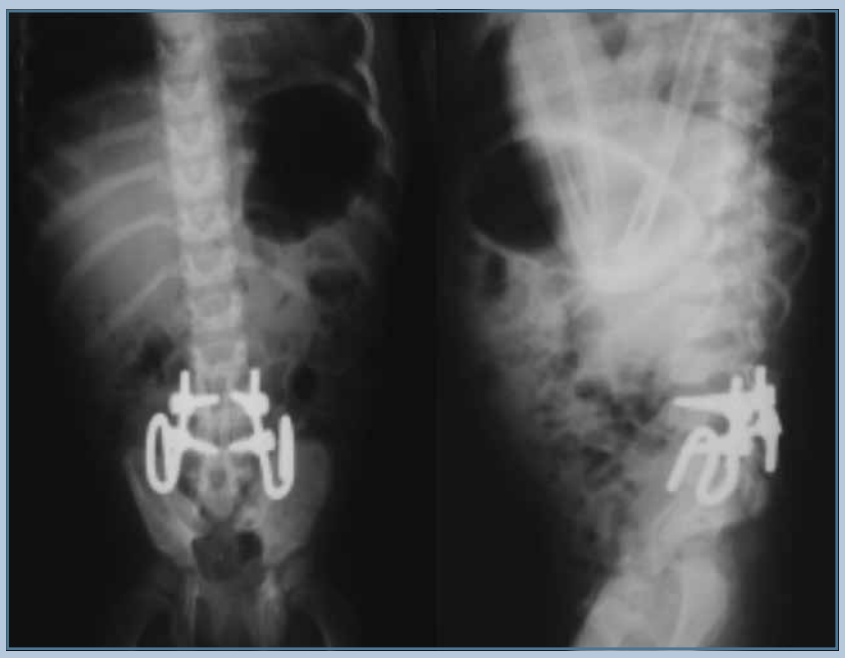

Fig. 8
X-ray picture of the patient S., 3 years old, after surgery

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