Complete Sacral Agenesis

—A Case Report—

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Summary: We report a case of a girl with complete sacral agenesis associated with arthrogrypotic-like deformity. This case demonstrates that a posterior knee release and braces following operation are effective treatment for knee-flexion contractures. Accordingly it is suggested that a supracondylar osteotomy is not required for mild or moderate recurrence of knee-flexion contractures.

Key words: complete sacral agenesis — lumbosacral agenesis — knee-flexion contractures — orthopaedic management — posterior knee release

Introduction

Complete sacral agenesis is a severe congenital malformation that is characterized by absence of the entire sacrum. It is often associated with arthrogryposis-like deformities of the lower extremity and malformations in the rectal area including an imperforate anus or rectal stenosis. All the patients had incontinence of urine and feces caused by spinal agenesis. Among orthopaedic problems in these patients, knee-flexion contractures with popliteal webbing were the most difficult to correct.

Case Report

This case is of a girl born at full term by Cesarean section in 1979. Both parents were nonconsanguineous and in good health. Her mother at the time of birth was 27 years old and her father 24. Her

mother had hydramnions and edema in late pregnancy. The birthweight was 2,750g. At birth it was noticed that the girl had severe knee-flexion contractures at 90-degrees flexion with externally rotated hip in the right lower limb, and bilateral club foot. Colostomy was performed two weeks after birth because of a developing megacolon and an imperforate anus.

Manual correction for the flexion-contractures of the hip, knee and feet was performed from 2 months to 2-and-a-half-years of age, but had no effect on the knee-flexion contractures. Mental development was normal and she could stand using her left leg only by holding onto something. She was first seen at our Orthopaedic Department at the age of 3-and-a-half years. On examination, the right lower extremity showed flexion-contractures with the hip at 25 degrees, 60-degrees in external rotation, and at 90-degree-knee flexion. The contracture

of the left knee was in flexion at 40 degrees with further flexion of 20 degrees (Fig. 1).

She had gibbus formation on her lower lumbar spine, a bilateral dimple in the groin, and a bilateral pes equinus. No motor function was present below the fourth lumbar vertebra. Sensation was normal except below the right knee. In manual muscle testing, the abdominal muscles were rated 4, hip flexors 3, and hip abductors and extensors were rated 0 bilaterally, and knee flexors 0 bilaterally, extensors 0 on the right side, and 3 on the left side. The anteroposterior X-ray of the pelvis revealed no sacrum with marked narrowing of the pelvis and no laminaformation below the second lumbar spine. From the third to the fifth vertebral bodies there was synarthrosis and connected fused ilia. The left hip was subluxated (Fig. 2).

When she was 4 years of age we performed a posterior capsulotomy on the right knee. A 7-cm-long-longitudinal incision was made in the popliteal fossa, but the location of the muscles could not be identified. The tendons of the medial and lateral side were dissected, but the flexion-contracture could not be corrected. Therefore, we made a lateral longitudinal skin incision so that all tendons and muscles were elongated and then performed a Z-plasty of skin webbing. Finally the passive knee extension could achieve 60 degrees and knee contracture was reduced to 30 degrees. A Quengel cast technique was used to extend the knee joint further. Simultaneously, we performed Achilles' tendon lengthening for the bilateral equinovarus deformities of the foot.

Two months after the operation, she could walk wearing long braces and using two crutches. We devised and produced a long brace with cuffs enabling her to be mobile. She was ambulatory freely with the braces and crutches. The flex-

Fig. 1. Photograph at 3 and a half years at admission shows arthrogryposis-like deformity in the right limb.

Fig. 2. Anteroposterior pelvic view shows no sacrum, synarthrosis from the 3rd to 5th vertebra, fused ilia and subluxation of the left hip.

Fig. 3. At the age of 5 years. Mild knee-flexion contractures were present, but she was a functional walker with bilateral long braces and crutches.

Fig. 4. At the age of 10 years. Recurrence of the flexion-knee contracture did not occur.

ion-contracture of the knee was only 15 degrees after operation (Fig. 3). At 10 years of age, she had flexion knee contractures of 45 degrees in the right knee, and 30 degrees with a further flexion of 15 degrees in the left knee. She could walk well with the long braces using crutches and she could even jump holding on crutches. There was no recurrence of the flexion-knee contractures (Fig. 4).

Discussion

Sacral agenesis is a rare condition consisting of partial or complete absence of the sacrum. Our case is classified as severe with total sacral agenesis and no lamina-formation below the second lumbar vertebra and with blocked vertebra

below the third vertebra. Renshaw (1978) has classified sacral agenesis into four types with the degree of severity increasing from type I through IV. Our case corresponds to a Renshaw type IV—variable lumbar and total sacral agenesis, with the caudal end-plate of the lowest vertebra resting above either fused ilia or an iliac amphiarthrosis. Phillips et al. (1982) have reported complete sacral agenesis combined with complete or partial lumbar deficiency as lumbosacral agenesis.

The incidence rate of sacral agenesis varies from 0.01 to 0.05 per 1000 live birth including patients with lumbosacral agenesis. A family history of a father and son has been reported, by Pouzet (1938). Rochet et al. (1966) have reported another case of familial partial agenesis in two

sisters with absence below the second sacral vertebra. Maternal diabetes may be a significant etiological factor. In the 192 cases described in the literature 16 diabetic mothers (8.33 per cent) have been reported (Dounis, 1978).

It has been further suggested that insulin injections may have a teratogenic effect on the embryo. Pederson et al. (1964) found in a study of 835 children of diabetic mothers that there was a 6.4% incidence rate of malformation and multiple congenital anomalies, six times more than in normal controls. In our case, the parents were in good health but her mother had hydramnions and edema in late pregnancy.

Of the orthopaedic problems of these patients, the knee flexion contractures are the most difficult to correct. It is said that the severity of the knee-flexion contractures is related to the quadriceps power; the weaker the quadriceps, the stronger the contractures. Our case had severe 90 degree knee-flexion contractures with hip-flexion contracture on one side.

It is also known that it is generally difficult to prevent the contractures from recurring. The posterior release and Z-plasty of webbing followed by wedging cast with Quengel's method exercise were successful in our case. After the operation, by wearing bilateral long braces with cuffs enabling mobility and walking actively with crutches, we managed to reduce the tendency to recurrent knee-flexion contractures. According to Banta and Nichols (1963), all patients had to undergo repeated anterior supracondylar closing wedge osteotomies because of resistant and recurrent knee flexion contractures. In the case described by Phillips et al. (1982), who had agenesis at the second lumbar level, there was an 80-degree knee-flexion contracture bilaterally. The first operation involved bila-

teral posterior capsulotomy at the age of four, after wedging casts, and contractures were reduced to 15 degrees. After four supracondylar osteotomies, she continued to have flexion contractures of 45 degrees in both knees up to the age of 27 years.

Our case demonstrates that a supracondylar osteotomy is not necessary to correct a moderate knee-flexion contracture. Our case had a 90-degree knee-flexion contracture and underwent a posterior capsulotomy at the age of four. Recurrence of the knee-flexion contracture of 15 degrees occurred 3 months after the operation. Since then the contracture did not increase for 6 years and she became a functionally good walker with moderate knee-flexion contracture.

The second orthopedic problem is spinal-pelvic instability. This instability was present in 9 of the 17 patients described by Andrich et al. (1979). Spinal-pelvic fusion has been advocated as an aid in correcting permanent flexion contractures of the hips. It is said that spinal-pelvic stabilization helps sitting balance and protects the viscera from compression. In our case spinal-pelvic instability was absent, so that comparatively easily she became an independent, functional walker.

It is not so difficult to correct the equinovarus deformities of the foot compared to the knee flexion contractures, and Achilles' tendon lengthening was effective. The mild hip-flexion contracture does not disturb orthotic fitting or the ability to walk. And the dislocation of the hip does not disturb walking, so that it has rarely been treated in childhood. If the patient has the acetabular dysplasia, subluxation or dislocation of the hip which cause hip pain after adolescence, a surgical treatment such as Chiari's osteotomy will be indicated.

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