Total Hip Reconstruction in a Woman With Cornelia de Lange Syndrome: A Case Report

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More than 250 cases of Cornelia de Lange syndrome have been reported in the medical literature, but none have described the use of hip reconstruction to correct the congenital dysplasia that may be associated with this condition. This article reports the application of a bipolar hemiarthroplasty and acetabular allograft reconstruction for a 32-year-old woman with congenital dysplasia and degenerative joint disease of the hip secondary to de Lange's syndrome. On admission, she was in considerable pain and unable to bear weight on the affected extremity. Her Harris hip score was 25. Following surgical intervention and a well-designed plan of rehabilitation, the patient's functional status improved markedly. Her Harris hip score was 72, and her pain was alleviated. (*J Natl Med Assoc.* 1997;89:530-532.)

Key words: Cornelia de Lange syndrome ◆ hip reconstruction ◆ typus degenerativus amstelodamensis

Cornelia de Lange syndrome or typus degenerativus amstelodamensis is a dysmorphogenic condition first described by the Dutch pediatrician Cornelia de Lange in 1933.[1] The essential characteristics of the syndrome are mental debility (IQ 40 to 85), hypertrichosis, brachymicrocephaly, hypertrophy of brows and lashes, mongoloid slant of eyes, depressed nasal bridge, thin broad lips with down-turned angles, and micrognathia. Other features commonly seen with the syndrome are micromelia often associated with oligodactylia, low-set ears, and syndactylism.[2-5] Because there are no definitive laboratory aids to confirm the diagnosis, the diagnosis is made primarily from the history and physical examination.[4,6,7]

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Of the more than 250 cases of Cornelia de Lange syndrome reported in the literature, none have involved the use of hip reconstruction to correct the congenital dysplasia occasionally associated with this condition. We believe this to be the first case report of the application of a bipolar hemiarthroplasty and acetabular allograft reconstruction for a patient with congenital dysplasia and degenerative joint disease of the hip secondary to Cornelia de Lange syndrome.

CASE REPORT

The patient was a 32-year-old woman with several congenital anomalies, including a right lower extremity terminal paraxial hemimelia of the tibia with absence of the great toe and first ray. At the age of 21 months, she underwent a right lower extremity amputation and conversion to a knee disarticulation amputation stump. She was fitted with a pylon-type plastic socket prosthesis, pelvic band, and SACH foot. By age 5½ years, she was able to ambulate independently without support, and her IQ was estimated to be <25. Speech was limited to a few words or phrases. Premorbid level of independent ambulation

was excellent. She was seen in the Handicapped and Crippled Children's Clinic at DC General Hospital until the age of 17, when she was lost to follow-up.

The patient presented again at the age of 32 in our clinic with complaints of progressive left hip pain. Family members reported that as her pain had increased, her ability to ambulate decreased. As the family noted, she was able to ambulate with only posterior support and assistance at the beginning of her reports of pain. Eventually, she was unable to bear weight on the lower left extremity and would only ambulate with excessive encouragement.

Physical examination revealed microcephaly and a low-riding hairline typical of Cornelia de Lange syndrome. Her eyebrows were bushy and extended across the midline, and her eyelashes were thick and curly. The pupils were normal and equally reactive to light. The facies showed many of the typical signs of Cornelia de Lange syndrome. The philtrum was long and prominent, the lips were thin, and the corners of the mouth were turned downward. The neck was short and mildly webbed. Marked hirsutism was evident on the forehead, neck, and sides of the face. A fine lanugo hair covered the back, shoulders, and extremities. Thoracic kyphoscoliosis was also evident.

An evaluation of the right upper extremity revealed hypomelia of the forearm and paraxial hemimelia of the ulna with only a single digit. Examination of the left upper extremity revealed a short humerus with terminal hemimelia of the ulna and two digits. The right and left elbows had flexion deformities of 130° and 110°, respectively. The third toe of the left foot was absent, but the lower left extremity was otherwise normal. The right lower extremity, which had been amputated above the knee, showed a well-healed stump. Physical examination of the left hip revealed that the patient had a 10° to 15° flexion contracture of the hip with flexion limited to 85°. Internal and external rotation were limited to approximately 10° to 15°. Her estimated Harris hip score was 25.

A radiographic analysis revealed that the left hip was dysplastic, with joint interval narrowing, subluxation, and a superior pseudoacetabulum. The femoral head appeared radiodense, and the superior half was distorted by dysplastic changes (Figure 1). A computed tomography scan of the left hip showed superior and lateral hip subluxation, sclerotic changes of the acetabulum and femoral head, and moderate joint effusion. Magnetic resonance imaging revealed increased signal activity on T2-weighted images, a finding consistent with osteonecrosis. Magnetic resonance imaging also revealed multiple subchondral cysts of the femoral head.

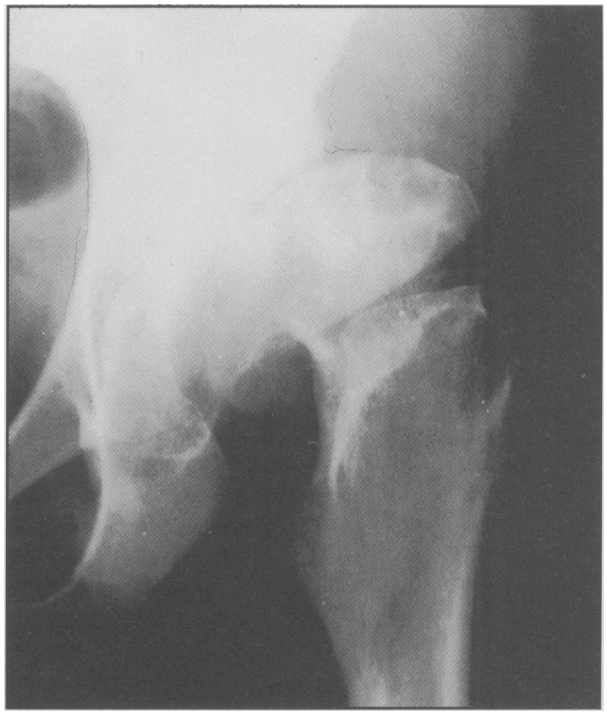

Figure 1.
Preoperative radiograph revealing a dysplastic left hip with lateral subluxation into a false acetabulum with osteophyte formation on the anterolateral surface of the acetabulum. In addition, there is evidence of degenerative joint disease involving the acetabulum and the femoral head.

Surgical intervention was considered appropriate based on these studies, the patient's declining ambulatory capacity, and consultations with the patient's family. A bipolar hip arthroplasty was determined to be the most appropriate procedure because of the patient's mental retardation and concern about hip dislocation.

In the fall of 1994, the patient underwent an elective bipolar hip arthroplasty with acetabular reconstruction with a superior dome allograft. The implant was an AML congenital dysplastic hip Bantom femoral stem 121 mm, noncemented and press-fit. The modular head was 28 mm in diameter with a +5-mm neck and a self-centered bipolar hip with a 52-mm outer diameter and a 28-mm inner diameter. Coxa breva and coxa magna were found

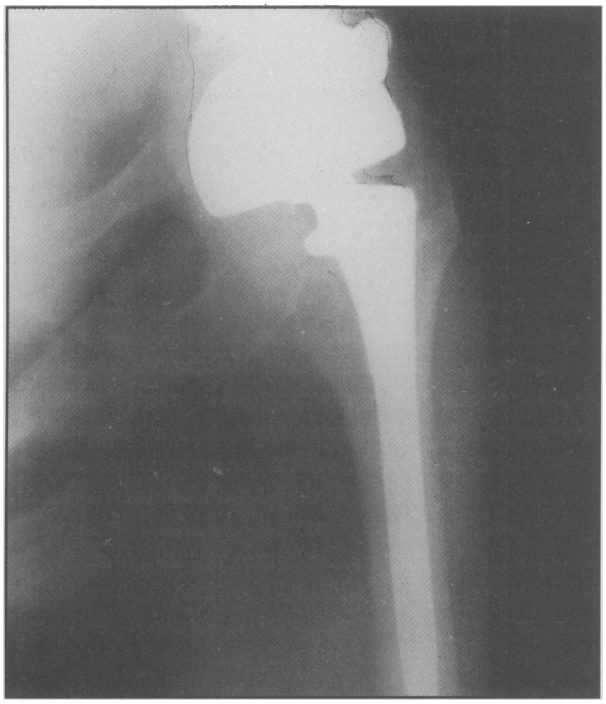

Figure 2.
Postoperative radiograph showing good placement of the femoral stem and bipolar acetabular component. The superior dome defect has been corrected by an allograft attached to the pelvis with three screws.

during surgery, with bony deficiencies of the posterior acetabulum and superior acetabular rim. The acetabular deficiencies were corrected with the processed femoral head and neck allograft attached to the pelvis using three cancellous screws. An allograft was preferred due to the poor bone quality of the patient's femoral head and neck. The partial osteotomy of the greater trochanter was repaired; two drains were inserted, one deep and one superficial to the tensor fascia lata.

Her postoperative course was complicated by an episode of aspiration pneumonia, which was treated with intravenous gentamicin. She also developed a postoperative superficial wound infection with a coagulase-negative *Staphylococcus*. She subsequently was given a 6-week course of vancomycin and the infection resolved completely.

Physical therapy was started postoperatively, and she began to ambulate with assistance. The patient was referred to a rehabilitation hospital for inpatient care. Rehabilitation progressed slowly, initially allowing the patient to ambulate with crutches, aided by her right lower extremity prosthesis. On follow-up visits, the patient was noted to be ambulating with minimal assistance approximately one to two blocks. The patient was able to climb stairs with minimal support. The range of motion in the left hip 10 weeks postsurgery revealed approximately 90° flexion, 20° abduction, 20° internal rotation, and 25° to 30° external rotation. Eventually, she was able to achieve a Harris hip score of 72. Radiographs showed the prosthesis in place with no evidence of subluxation or dislocation (Figure 2).

DISCUSSION

Our patient presented with many of the physical characteristics of Cornelia de Lange's syndrome. She was severely mentally retarded and had severe disability and pain. Surgical intervention appeared to be the best means of relieving the pain, restoring functional capacity to the hip, and allowing her some measure of independence.

This case demonstrates that appropriate and timely surgical intervention can produce good results in patients with dysplastic hips associated with congenital anomalies. Considering her extensive physical and mental disabilities, our patient attained a remarkably good postoperative Harris hip score (72, from a preoperative score of 25) and ambulatory capability.

Careful postoperative management and rehabilitation are necessary in patients with such severe disabilities. A diligent rehabilitation program must be instituted to ensure adequate range of motion and ambulatory function.

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