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Total Hip Arthroplasty Reduces Pain and Improves Function in Patients With Spondyloepiphyseal Dysplasia: A Long-Term Outcome Study of 50 Cases

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Abstract

Background: Spondyloepiphyseal dysplasia (SED) is rare genetic condition which leads to skeletal and joint deformities that can predispose patients to degenerative joint disease. There are limited reports on the results of total hip arthroplasty (THA) in this patient population. The purpose of this study is to review clinical and radiographic outcomes of THA performed in patients with SED at one institution.

Methods: Among 43,917 patients undergoing primary THA from 1970 to 2015, we identified 50 THAs performed in 29 patients with SED; 21 patients underwent bilateral THA (none simultaneous). There were 16 females and 13 males; mean age, body mass index, and height were 39 years, 28.7 kg/m², and 145 cm, respectively. All patients were able to ambulate prior to the THA. Mean follow-up was 11 years (range 2–38).

Results: Mean implant survival for primary THA in SED patients at the 5, 10, and 20-year time points was 96%, 85%, and 55%, respectively. Thirteen patients required revision THA, most commonly for polyethylene wear (n = 6) and aseptic loosening (n = 5), and 4 additional patients underwent nonrevision reoperations. Prior to surgery, 90% of patients had severe or moderate pain, which was reduced to 8% of patients postoperatively ($P < .001$). Mean Harris Hip Score improved from 47 to 87 ($P < .001$). Prior to surgery, 64% of patients required gait aids, which reduced to 34% postoperatively ($P < .001$).

Conclusion: THA provided significant pain reduction and improvement in function, with a majority of patients ambulating independently following the procedure. There was a high incidence of complications following THA in patients with SED, most commonly secondary to polyethylene wear and osteolysis from conventional polyethylene and historical implants.

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Keywords

clinical outcomes; spondyloepiphyseal dysplasia; total hip arthroplasty; THA; Complications; skeletal dysplasia

Spondyloepiphyseal dysplasia (SED) is a form of short-trunk dwarfism caused by epiphyseal defects secondary to abnormal synthesis of Type II collagen [1]. Premature osteoarthritis, particularly of the hip joint, is a common sequelae of SED and related skeletal dysplasias as a result of abnormal bone and joint development in conjunction with altered biomechanics [2]. Patients with SED frequently have complex hip deformities with coxa vara being a hallmark of disease. These deformities can be addressed with a variety of hip preservation techniques including osteotomies of the proximal femur and/or acetabulum as well as soft tissue releases [3]. Despite these attempts at joint salvage, many patients with SED develop premature coxarthrosis [4].

Although total hip arthroplasty (THA) has become a straight-forward procedure for primary osteoarthritis, patients with SED and related skeletal dysplasias have unique anatomic considerations that present additional technical challenges [5]. The existing literature is limited by small cohorts, short-term follow-up, or heterogeneous series evaluating multiple forms of dwarfism and skeletal dysplasia. The purpose of this study is to review the long-term clinical and radiographic outcomes of THA performed in patients with SED at one institution with a focus on need for revision, reoperation, and postoperative complications and how these outcomes have changed in the face of implant evolution.

Methods

Following Institutional Review Board approval, our Total Joint Registry was utilized to review the medical records of all patients undergoing primary THA from 1970 to 2015. Patients with a diagnosis of SED were included in this study and none were excluded.

A standard protocol was used to obtain clinical and radiographic follow-up at 3 months, 6 months, 1 year, 2 years, and every 5 years after THA. All patients undergoing total joint arthroplasty at our institution were evaluated in person with a modified Harris Hip Score (HHS) or completed a standardized questionnaire at every follow-up visit; these questionnaires were used to assess modified HHSs preoperatively and postoperatively [6]. Radiographs were obtained at these same time points including an anteroposterior pelvis, anteroposterior hip, and lateral of the hip.

The Kaplan-Meier method was used to establish survivorship curves at 5, 10, 20, and 30 years [7]. Two authors assembled revision, reoperation, complication, and nonhip orthopedic procedural data using electronic medical records and diagnostic codes (C.C.W. and J.A.P.). The difference in HHS was determined by comparing preoperative scores to data obtained at the latest follow-up visit for all patients; those who experienced death, revision, or reoperation were censored at the visit before the event. All statistical tests were 2-sided and a P value $<.05$ was considered significant.

Results

A search of our Total Joint Registry returned data on 43,917 patients undergoing primary THA performed over a 46-year period (1970–2015). The inclusion and exclusion criteria were applied by a single author (C.C.W.) to yield a cohort of 50 THAs (0.11%) performed in 29 patients with SED. Mean follow-up was 11 years (range 2–38).

There were 16 females and 13 males. Mean age, body mass index, and height were 39 years (range 16–79), 28.7 kg/m² (range 15.5–56.9), and 145 cm (range 93–179), respectively (Table 1). All patients were able to ambulate prior to THA (Table 1). Among the 29 patients evaluated, 13 (44%) had a diagnosis of SED confirmed by a medical geneticist at our institution, with the remainder of diagnoses assigned clinically by a combination of orthopedic surgeons, pediatricians, and related subspecialists (Table 1). Twenty-one patients underwent bilateral THA, representing 42 cases (84%). None of these patients received bilateral THA under a single anesthetic.

Before THA, 12 patients (24%) had at least 1 surgical procedure on the ipsilateral hip (range 1–2). Six patients (12%) underwent proximal femoral osteotomy, 4 patients (8%) underwent interposition arthroplasty, and 3 patients (6%) underwent a pelvic osteotomy. All THAs were performed by experienced reconstructive surgeons at a single institution. The surgical approach was posterolateral in 23 cases (46%), transtrochanteric in 16 cases (32%), and anterolateral in 11 cases (22%). THA fixation was uncemented in 32 cases (64%), hybrid in 11 cases (22%), cemented in 6 cases (12%), and reverse hybrid in 1 case (2%). A constrained acetabular component was used in 1 case. Bearing surface combinations were metal-on-polyethylene in 40 cases (80%), ceramic-on-polyethylene in 6 cases (12%), and ceramic-on-ceramic in 4 cases (8%). Among the 46 cases that used polyethylene, highly cross-linked polyethylene (HXPLE) was used in 24 cases (52%) and conventional polyethylene was used in the remaining 22 cases (48%).

Mean implant survival for primary THA in SED patients at the 5, 10, and 20-year time points was 96%, 85%, and 55%, respectively (Fig. 1). Among the 24 cases that received HXPLE, there was 1 failure at 2-year follow-up due to aseptic loosening of the femoral component, whereas the other 23 cases (96%) had intact primary implants at mean final follow-up of 6 years (range 2–14, $P < .001$) (Fig. 2).

Thirteen cases (26%) required component revision surgery as follows: both components ($n = 3$), acetabular component ($n = 3$), femoral component ($n = 5$), and head and liner exchange ($n = 7$) (Table 2). The most common indications for revision THA was polyethylene wear ($n = 6$)—which occurred in 4 hybrid THAs and 2 uncemented THAs—and aseptic loosening ($n = 5$), which occurred in 2 hybrid THAs and 3 uncemented THAs (Table 3). Three patients were revised for periprosthetic fracture and another for periprosthetic infection (Tables 2 and 3). In addition to revision procedures, 4 patients underwent nonrevision reoperation: 3 to remove painful hardware and 1 for irrigation and debridement (Table 2). Twelve of the 13 revisions (92%) and 2 of the 4 reoperations (50%) were performed in patients with conventional polyethylene (Table 3). A complete list of postoperative complications is provided in Table 4. Twelve patients went on to undergo additional nonhip orthopedic

surgical procedures (Table 5). This subset of patients did not differ significantly from the study cohort with respect to age at index THA, height, or body mass index.

Prior to surgery, 90% of patients had severe or moderate pain, which decreased to 8% postoperatively ($P < .001$). Mean preoperative HHS was 47, improving to 87 postoperatively ($P < .001$). Prior to surgery, 64% of patients required use of a gait aid which decreased to 34% postoperatively ($P < .001$) (Table 1). The minimal clinically important improvement threshold in HHS score at 5 years has been estimated at 16–18 points [8]. For a sample size of 50 cases with an observed standard deviation of 13.56, we can detect a 16-point difference in HHS score with power equal to 0.98.

Discussion

There are few reports in the literature evaluating the long-term outcomes of THA in patients with SED; therefore, the purpose of this investigation is to report outcomes of THA in 50 patients with SED at our institution. THA was effective in reducing pain and improving function, with a majority of patients ambulating independent of gait aids following the procedure (Fig. 3). The revision, reoperation, and complication rates were high by 20 years in this case series of mostly young patients. However, most poor outcomes were secondary to the use of conventional polyethylene and historical implants, suggesting that modern materials, implants, and surgical techniques render THA a reliable option for end-stage coxarthrosis in SED patients (Fig. 4).

A previous study from our institution reported on 62 THAs performed between 1971 and 1997 in a group of patients with dwarfism and some form of osteochondrodysplasia [5]. They observed a higher rate of revision (30%); however, the indications for revision were similar to our cohort with aseptic loosening being most common in addition to a few periprosthetic fractures and infections. Notably, all these procedures were performed with conventional polyethylene and one or both components were cemented in 71%. Despite many failures, they demonstrated improvement in pain and function for most patients. A more contemporary cohort of 102 patients with dwarfism undergoing THA was analyzed by Modi et al [9]. Short- to mid-term outcomes were analyzed and compared to a matched cohort of normal stature. They showed comparable survivorship outcomes between groups at the 2, 5, and 10-year time points. There was a mild trend toward increased periprosthetic fractures and wear-related failures. Another recent report by Guenther et al [10] evaluated short-term 1 and 5-year outcomes of a group of 127 patients with dwarfism undergoing THA. The overall implant-associated survival rate after 1 year of follow-up was 99.1% and after 5 years of follow-up was 92.6%, which is slightly lower than survivorship rates from historical reports in normal stature individuals from their practice, although no formal matching was performed.

The most common indication for revision in our cohort was aseptic loosening, of which half of the cases were comprised of uncemented implants. Reports of aseptic loosening in similar cohorts have been variable. Lim et al [11] reported no revisions due to acetabular or femoral loosening in a cohort of 23 THAs in patients with multiple epiphyseal dysplasia at a mean follow-up of 4.8 years. The incidence of aseptic loosening of the uncemented acetabular

component as an indication for revision in a cohort of 16 THAs in 9 patients with multiple epiphyseal dysplasia was 30% at a mean follow-up of 12.5 years postoperatively [12]. In a population-based study of 42,665 primary THAs from the New Zealand Joint Registry, the rate of revision for aseptic loosening of the acetabular component was greater in fully cemented groups, compared to uncemented and hybrid fixation techniques; the rate of revision of femoral components was similar between cemented and uncemented groups for patients under 75 years of age [13].

The current study and selected reports on similar populations with skeletal dysplasia show that results of THA approach success rates seen in the general population with modern implants. The majority of failures in studies including patients who received THA before the advent of HXPLE show elevated failure rates above historical reports of THA during the same time period. Complications associated with conventional polyethylene, cement fixation, and implants with fewer options for modularity likely had a greater negative impact on patients with SED and related skeletal dysplasias. This population typically is younger in addition to having a collection of anatomic and biomechanical considerations that historic implants and techniques were not equipped to handle. Many authors have demonstrated that HXLPE exhibits lower linear and volumetric wear rates, as well as a decreased rate of periprosthetic osteolysis, than older, conventional components [14–17]. We hypothesize that contemporary implants with HXLPE, improved ingrowth surfaces, and increased modularity will continue to demonstrate decreased osteolysis and failure rates due to aseptic loosening; however, longitudinal study of the more recent cases in this series will be mandatory to confirm that premise.

This study must be interpreted in light of important limitations. First, it is a small, retrospective case series from a single institution and is therefore subject to the limitations inherent to that study design. Second, due to the small numbers, we were unable to evaluate risk factors for revision, aside from polyethylene type, which is further confounded by the fact that conventional polyethylene was most often present with cemented components and historical implants. There remains value in the present report, however, as it is the largest clinical series to our knowledge of patients with SED treated with THA and one of the few with long-term outcome data for any type of skeletal dysplasia.

SED and related skeletal dysplasias commonly require management of debilitating coxarthrosis at a younger age than primary osteoarthritis. There are numerous challenges to performing THA in this population secondary to short stature, abnormal skeletal morphology about the hip joint, and prior surgery for hip preservation. The current study demonstrates that despite these technical considerations, THA provides predictable improvement in pain and function with maintenance of independence. Complications, revisions, and reoperations were high in this series; however, the vast majority was related to use of conventional polyethylene and historical implants with fewer options to accommodate SED anatomy. Modern implants with HXPLE and modularity or custom design when necessary render THA a reliable option for patients with SED suffering from symptomatic coxarthrosis.

References

- [1]. Terhal PA, Nievelstein RJ, Verver EJ, Topsakal V, van Dommelen P, Hoornaert K, et al. A study of the clinical and radiological features in a cohort of 93 patients with a COL2A1 mutation causing spondyloepiphyseal dysplasia congenita or a related phenotype. *Am J Med Genet A* 2015;167A: 461–75. [PubMed: 25604898]
- [2]. Kopits SE. Orthopedic complications of dwarfism. *Clin Orthop Relat Res* 1976;114:153–79.
- [3]. Bayhan IA, Abousamra O, Rogers KJ, Bober MB, Miller F, Mackenzie WG. Valgus hip osteotomy in children with spondyloepiphyseal dysplasia congenita: midterm results. *J Pediatr Orthop* 2017 10.1097/BPO.0000000000000945 [Epub ahead of print].
- [4]. Treble NJ, Jensen FO, Bankier A, Rogers JG, Cole WG. Development of the hip in multiple epiphyseal dysplasia. Natural history and susceptibility to premature osteoarthritis. *J Bone Joint Surg Br* 1990;72:1061–4. [PubMed: 2246289]
- [5]. Chiavetta JB, Parvizi J, Shaughnessy WJ, Cabanela ME. Total hip arthroplasty in patients with dwarfism. *J Bone Joint Surg Am* 2004;86-A:298–304.
- [6]. Harris WH. Traumatic arthritis of the hip after dislocation and acetabular fractures: treatment by mold arthroplasty. An end-result study using a new method of result evaluation. *J Bone Joint Surg Am* 1969;51:737–55. [PubMed: 5783851]
- [7]. Goel MK, Khanna P, Kishore J. Understanding survival analysis: Kaplan-Meier estimate. *Int J Ayurveda Res* 2010;1:274–8. [PubMed: 21455458]
- [8]. Singh JA, Schleck C, Harmsen S, Lewallen D. Clinically important improvement thresholds for Harris Hip Score and its ability to predict revision risk after primary total hip arthroplasty. *BMC Musculoskelet Disord* 2016;17:256. [PubMed: 27286675]
- [9]. Modi RM, Kheir MM, Tan TL, Penny GS, Chen CL, Shao H, et al. Survivorship and complications of total hip arthroplasty in patients with dwarfism. *Hip Int* 2017;27:460–4. [PubMed: 28497456]
- [10]. Guenther D, Kendoff D, Omar M, Cui LR, Gehrke T, Haasper C. Total hip arthroplasty in patients with skeletal dysplasia. *J Arthroplasty* 2015;30: 1574–6. [PubMed: 25882609]
- [11]. Lim SJ, Park YS, Moon YW, Jung SM, Ha HC, Seo JG. Modular cementless total hip arthroplasty for multiple epiphyseal dysplasia. *J Arthroplasty* 2009;24: 77–82.
- [12]. Ramaswamy R, Kosashvili Y, Cameron H. Total hip replacement in patients with multiple epiphyseal dysplasia with a mean follow-up of 15 years and survival analysis. *J Bone Joint Surg Br* 2010;92:489–95. [PubMed: 20357323]
- [13]. Hooper GJ, Rothwell AG, Stringer M, Frampton C. Revision following cemented and uncemented primary total hip replacement: a seven-year analysis from the New Zealand Joint Registry. *J Bone Joint Surg Br* 2009;91:451–8. [PubMed: 19336803]
- [14]. Callary SA, Field JR, Campbell DG. Low wear of a second-generation highly crosslinked polyethylene liner: a 5-year radiostereometric analysis study. *Clin Orthop Relat Res* 2013;471:3596–600. [PubMed: 23893361]
- [15]. de Steiger RN, Muratoglu O, Lorimer M, Cuthbert AR, Graves SE. Lower prosthesis-specific 10-year revision rate with crosslinked than with non-crosslinked polyethylene in primary total knee arthroplasty. *Acta Orthop* 2015;86:721–7. [PubMed: 26119884]
- [16]. Glyn-Jones S, Thomas GE, Garfjeld-Roberts P, Gundle R, Taylor A, McLardy-Smith P, et al. The John Charnley Award: highly crosslinked polyethylene in total hip arthroplasty decreases long-term wear: a double-blind randomized trial. *Clin Orthop Relat Res* 2015;473:432–8. [PubMed: 25123239]
- [17]. Teeter MG, Yuan X, Somerville LE, MacDonald SJ, McCalden RW, Naudie DD. Thirteen-year wear rate comparison of highly crosslinked and conventional polyethylene in total hip arthroplasty: long-term follow-up of a prospective randomized controlled trial. *Can J Surg* 2017;60:212–6. [PubMed: 28570216]

Fig. 1. Survivorship of THA in patients with SED: Kaplan-Meier curve demonstrating THA implant survivorship in patients with SED. The 5, 10, and 20-year survivorship was 96%, 85%, and 55%, respectively.

Fig. 2.

Survivorship of THA in patients with SED by implant: Kaplan-Meier curve demonstrating THA survivorship by implant. The 5 and 10-year survivorship for HXPLE was 96%. Conventional polyethylene survivorship was 96%, 82%, and 58% at 5, 10, and 20 years, respectively. HXPLE, highly cross-linked polyethylene; PLE, conventional polyethylene.

Fig. 3. Bilateral THA with 2 modalities of failure secondary to conventional polyethylene wear. (A) Two-year postoperative anteroposterior pelvis radiograph of a patient with SED who received bilateral hybrid THA with conventional polyethylene. (B) Twelve-year postoperative anteroposterior pelvis radiograph showing a Vancouver B2 periprosthetic fracture of the left hip secondary to a ground level fall. There is eccentric polyethylene wear and significant osteolysis around the femoral and acetabular components. The right hip demonstrates eccentric polyethylene wear and acetabular osteolysis in the setting of a symptomatic hip. The left hip was revised at the time of fracture with head and liner exchange and placement of a modular fluted tapered femoral component. The right hip was revised 7 months later with head and polyethylene liner exchange in addition to retroacetabular bone grafting.

Fig. 4. Successful THA using modern implants and highly cross-linked polyethylene in a patient with SED. (A) Preoperative AP pelvis radiograph of a patient with SED. (B) Postoperative AP pelvis radiograph at 10-year follow-up showing well-fixed and appropriately positioned bilateral THA in an asymptomatic patient.

Table 1

Characteristics of Patients With Spondyloepiphyseal Dysplasia Undergoing THA.

Demographic	Mean (Range)
Age (y)	39 (16–79)
Height (cm)	145 (93–179)
BMI (kg/m ²)	28.7 (15.5–56.9)
Gender (male:female)	13:16
Diagnosis	Count (%)
Spondyloepiphyseal dysplasia	29 (100)
Confirmed by geneticist	13 (45)
Harris Hip Scores	Mean (range)
Mean preoperative (range)	47 (24–72)
Mean postoperative (range)	87 (39–100)
Ambulatory status	Count (%)
Preoperative ambulator	50 (100)
Preoperative gait aids	32 (64)
Postoperative gait aids	17 (34)

BMI, body mass index; THA, total hip arthroplasty.

Table 2

Revision and Reoperation Counts for All Patients Following THA.

Revision/Reoperation—Overall	Count
Revision surgery indication	
Aseptic loosening	5
Polyethylene wear	6
Periprosthetic infection	1
Periprosthetic fracture	3
Recurrent instability	2
Component revised	
Both components	3
Acetabular component	3
Femoral component	5
Head and liner exchange	7
Nonrevision reoperation	
Removal of hardware	3
Irrigation and debridement	1

THA, total hip arthroplasty.

Table 3

Indications for Revision THA by Fixation and Bearing Surface.

Revisions by Construct	Count (%)
Bearing surface	
Metal-on-conventional PE	12 (92)
Metal-on-HXPLE	1 (8)
Fixation	
Hybrid	5 (38)
Aseptic loosening	2
Polyethylene wear	4
Periprosthetic infection	0
Periprosthetic fracture	1
Recurrent instability	0
Cemented	3 (24)
Aseptic loosening	0
Polyethylene wear	0
Periprosthetic infection	1
Periprosthetic fracture	2
Recurrent instability	0
Uncemented	5 (38)
Aseptic loosening	3
Polyethylene wear	2
Periprosthetic infection	0
Periprosthetic fracture	0
Recurrent instability	2

THA, total hip arthroplasty; PE, polyethylene; HXPLE, highly cross linked polyethylene.

Table 4

Complications Following THA.

Complications	Count(%)
Postoperative complications	21 (42)
Polyethylene wear	8 (16)
Femoral osteolysis	4 (8)
Acetabular osteolysis	4 (8)
Periprosthetic fracture	1 (2)
Symptomatic DVT	1 (2)
Heterotopic ossification	1 (2)
Deep infection	1 (2)
Superficial infection	1 (2)

THA, total hip arthroplasty; DVT, deep vein thrombosis.

Table 5**Nonhip Orthopedic Procedures After Index THA.**

Demographic	Mean (Range)
Age (y)	40 (18–62)
Height (cm)	138 (93–157)
BMI (kg/m ²)	28.0 (19.9–42.9)
Gender (male:female)	4:8
Spine	Count (%)
Partial Harrington rod removal	1 (25)
C2 laminectomy; posterior occipital-C2 fusion	1 (25)
Decompressive laminectomy: T3-T7	1 (25)
Reoperation laminectomies: T3–4, T5–6	1 (25)
Lower extremity	Count (%)
Total knee arthroplasty	11 (85)
Ankle arthrodesis	1 (7.5)
Transmetatarsal amputation	1 (7.5)
Upper extremity	Count (%)
Total shoulder arthroplasty	2 (50)
Total elbow arthroplasty	2 (50)

BMI, body mass index; THA, total hip arthroplasty.