# BILATERAL TOTAL HIP REPLACEMENT IN PSEUDOACHONDROPLASIA

WIRTZ D.C., BIRNBAUM K., SIEBERT C.H., HELLER K.D.

**Pseudoachondroplasia is an inherited skeletal dysplasia with short-limbed dwarfism and early onset of osteoarthritis. A 29-year-old pseudoachondroplastic woman presented with progressively painful hips secondary to severe osteoarthritis of both joints, so that total joint replacements were necessary to restore her mobility and quality of life. The implants inserted had to be specifically manufactured in accordance with the individual geometry and reduced bone size. In addition, the implants mechanical resistance to dynamic loading conditions had to be tested prior to total hip replacement surgery.**

**Key words :** pseudoachondroplasia ; hypochondroplasia ; total hip replacement ; osteoarthritis.
**Mots-clés :** pseudoachondroplasie ; hypochondroplasie ; prothèse totale de hanche ; arthrose.

## INTRODUCTION

Pseudoachondroplasia is an autosomal dominant dwarfing condition associated with disproportionate short stature and marked joint deformities. In contrast to classic achondroplasia, no typical changes of the cranium are found. The disease genes in families with pseudoachondroplasia have been localized on the short arm of chromosome 19 (1,3), but most cases result from a spontaneous mutation without a family predisposition. The pathological cause of the restricted growth is seen in a disturbance of endochondral ossification and inhibition of cartilage proliferation. The early onset of osteoarthritis is one of the main clinical problems of individuals with pseudoachondroplasia. According to the literature, the degenerative changes become symptomatic in the fifth decade of life and most frequently affect the hip joint (6). In the case presented here, a young woman with pseudoachondroplasia suffered from severe osteoarthritis of both hips, requiring total hip replacement surgery to restore her mobility.

## CASE REPORT

A 29-year-old woman, 122 cm in height and 46 kg weight, presented with severe pain in both hips with marked progression over the previous 18 months. The maximum walking distance was reduced to 500 meters, and pain-free weight bearing was no longer possible.

The phenotypical features were characterized by disproportionate short stature with micromelia, relative macrocephaly, genua valga and increased lumbar lordosis (fig.1a, 1b). There was neither mental retardation nor neurological involvement. Clinically, the movement of both hips was painfully restricted, especially in abduction and internal rotation. The left lower limb was 1.5 cm shorter than the right. The Trendelenburg test was positive bilaterally. Radiologically, a slightly reduced interpedicular distance of the lumbar spine with isthmic spondylolysis L4-L5 and L5-S1 was found. The radiograph of the pelvis revealed squared and shortened iliac bones with dysplastic acetabular roofs and marked osteoarthritic destruction of the femoral head on both sides (fig. 2).

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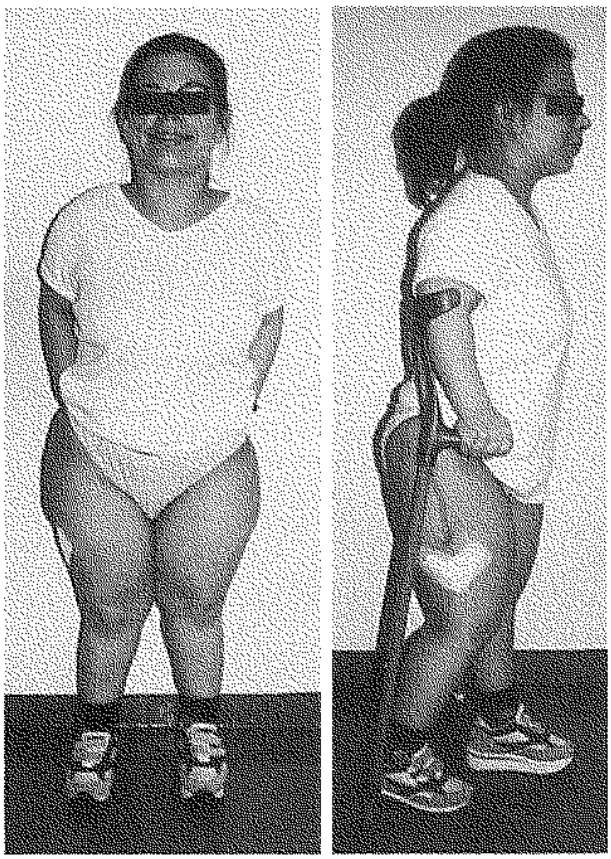

*Fig.1a/b.* — Frontal and lateral photograph of a 29-year-old woman after total hip arthroplasty on the right side: short-limbed dwarfism, 1.5-cm leg length discrepancy corrected by shoe modification on the left, slight genua valga, normal craniofacial appearance and normal hand configuration.

Preoperatively, both proximal femurs were scanned by CT (Somatom Plus, Siemens, Erlangen, Germany) to calculate the minimum transverse diameters of the intramedullary canal. Based on these data, a customized femoral stem was manufactured by Aesculap AG & CO.KG (Tuttlingen, Germany), in accordance with the material-related standards of ISO-norm 7206. Breaking stress computations and ultimate strength tests revealed that only a cemented stem design based on a CoCrMo-alloy could guarantee reliable mechanical resistance to dynamic loading conditions. Titanium-based alloys for cement-free implantation showed insufficient structural stability under the required geometric conditions.

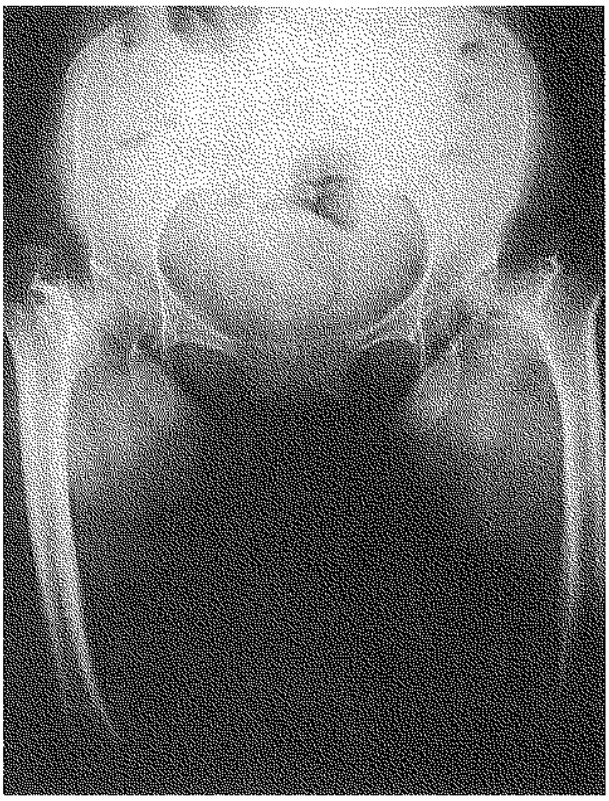

*Fig.2.* — Preoperative anteroposterior radiograph of the pelvis: dimished flare of both iliac bones with acetabular dysplasia and coxa planovara with osteoarthritic destruction of the femoral heads.

With an interval of four months between the procedures, both hips were replaced using the customized femoral implants. They were fixed with Refobacin Palacos (Merck, Darmstadt, Germany) and modern cementation technique (vacuum mixing, pulsatile lavage, distal femoral plug, stem centralizer, retrograde pressurized filling). On the acetabular side, an additional autogenous femoral head graft was necessary to reconstruct the dysplastic acetabular roof prior to implantation of standard cemented polyethylene cups (fig. 3). Because of the abnormal shape of the anterior and posterior columns of the acetabulum, a cementless cup implantation was not possible.

Postoperatively, both legs were identical in length, and after rehabilitation the patient was able to ambulate freely without assistance. The follow-up time is now 18 months on the left side and 14

months on the right side. Both hips are functioning well with a painfree and unlimited walking ability of the patient. Radiographically, there are no signs of prostheses loosening with adequate osseous integration of the autogenous grafts as structural support of the dysplastic acetabula.

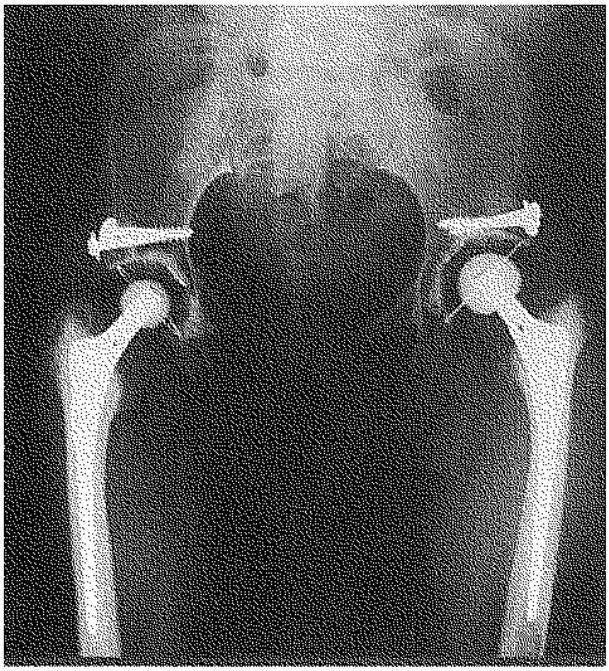

*Fig.3.* — Postoperative situation with cemented total hip arthroplasties on both sides. *Left:* cemented CoCrMo-alloy stem (BiContact®, Aesculap AG, Tuttlingen, Germany) with tapered sleeve 8/10, 28-mm aluminum oxide ceramic head, 46-mm cemented polyethylene cup, acetabular augmentation with autogenous graft. *Right:* cemented CoCrMo-alloy stem (BiContact®, Aesculap AG, Tuttlingen, Germany) with tapered sleeve 8/10, 22-mm aluminum oxide ceramic head, 42-mm cemented polyethylene cup, autogenous graft as structural support of the dysplastic acetabulum.

## DISCUSSION

Pseudoachondroplasia is a well-characterized autosomal dominant dwarfing condition with spondyloepimetaphyseal dysplasia. Previous genetic studies provided mapping of the pseudoachondroplasia gene to chromosome 19, which codes for a noncollagenous protein expression in the territorial matrix of chondrocytes (2). Clinically, the syndrome is characterized by disproportionate short stature, marked joint deformities, and early-onset osteoarthritis. In contrast with achondroplasia, the growth disturbance is not recognizable at birth. The shortening of the extremities develops mainly during childhood, while typical changes of the face and cranium are missing (5,7).

A problem in diagnosing pseudoachondroplasia is that the pathognomonic changes of vertebral bodies invariably disappear around the age of 10. An adult might subsequently be misdiagnosed as suffering from multiple epiphyseal dysplasia, Fairbank type, even though the deformities are less severe than in pseudoachondroplasia (4). A knowledge of the spectrum of clinical features of pseudoachondroplasia is therefore paramount in reaching the correct diagnosis.

As documented in this case, individuals suffering from pseudoachondroplasia can develop extreme joint deformities, especially in the hips. In young adult pseudoachondroplastic patients, the pelvic changes are frequently minimal and characterized by diminished flare of iliac bones and short femoral necks. The severe osteoarthritis with marked coxa planovara usually develops during the later course of life and is the major cause of discomfort in the older adult population (6). Even though severe osteoarthritic destruction of the hip joint is rare in younger affected patients, total hip replacement seems justified in all cases with clinical symptoms, reduced mobility and typical radiological signs of premature osteoarthritis. Femoral head preserving methods are generally not indicated because the osteoarthritic changes are primarily concentric.

Because of the reduced size of bony structures in pseudoachondroplastic patients, an exact preoperative evaluation with geometric analysis including CT-scan of the proximal femur and acetabulum is necessary prior to performing total hip replacement surgery. The implants must be adapted to the individual situation of the patient. Reliable mechanical resistance to dynamic loading has to be taken into account when manufacturing such implants. Breaking stress computations and ultimate strength tests must be carried out in accordance with the material-related standards of ISO-norm 7206. In addition, the wear problem of polyethylene cups must be taken into account when using small cups

with low diameters. To avoid increased stresses in the polyethylene with an unacceptable risk of premature wear the standard midrange head size of 28-mm must be reduced to 22-mm components.
In summary, total hip replacement is justified in young adult pseudoachondroplastic patients suffering from marked premature osteoarthritis of the hip. The improved range of motion of the affected joint, increased mobility and quality of life are encouraging. The required implants should be manufactured individually, respecting the specific geometric and material-related conditions.

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## RÉSUMÉ

*WIRTZ D.C., BIRNBAUM K., SIEBERT C.H., HELLER K.D. Remplacement bilatéral de la hanche par prothèse totale dans un cas de pseudo-achondroplasie*

La pseudo-achondroplasie est une dysplasie génétiquement transmissible. La dysplasie est associée à un nanisme et une tendance à l'arthrose précoce. Les auteurs rapportent le cas d'une femme qui présentait une arthrose grave des deux hanches, imposant un remplacement bilatéral par prothèse totale. Les prothèses ont dû être fabriquées sur mesure, en fonction de la géométrie individuelle des os et de leurs dimensions réduites. De plus, la résistance mécanique des prothèses a dû être testée avant l'implantation.

## SAMENVATTING

*WIRTZ D.C., BIRNBAUM K., SIEBERT C.H., HELLER K.D. Bilaterale heupvervangingsartroplastiek in een geval van pseudo-achondroplasie.*

Pseudo-achondroplasie is een erfelijke dysplasie, onder meer gekenmerkt door dwerggroei en neiging tot vroegtijdige artrose. De auteurs beschrijven het geval van een vrouw met ernstige artrose van beide heupen, waarvoor bilaterale vervangingsartroplastiek noodzakelijk was. De prothesen moesten op maat gemaakt worden, in functie van de aparte geometrie en van de geringe afmetingen van de beenderen. Daarenboven was het nodig vooraf de mechanische weerstand van de prothesen te testen.