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Joint replacement associated with ochronosis in three cases, two were siblings

Okronozis ile ilişkili eklem replasmanı yapılan üç olgu, ikisi ikiz

Mustafa Deveci[1], Alper Çıraklı[1], Erdal Uzun[1], Eyyüp Sabri Öncel[2]

[1]Department of Orthopaedics and Traumatology, Kayseri Research and Training Hospital, Kayseri,Turkey
[2]Department of Orthopaedics and Traumatology, Batman State Hospital, Batman, Turkey
Corresponding author: Mustafa Deveci, Department of Orthopaedics and Traumatology, Kayseri Research and Training Hospital, Kayseri,Turkey
E-mail: drmdeveci@gmail.com
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SUMMARY

Homogentisic acid which accumulates due to homogentisic acid oxidase enzyme deficiency causes joint cartilage damage which then, together with ageing causes osteoarthritis. In this paper, the outcomes are evaluated of arthroplasty applied because of ochronosis-related advanced stage osteoarthritis. The study included 6 joints of 3 cases in which ochronosis-related advanced stage osteoarthritis developed and arthroplasty was applied between 2000 and 2015. The cases were 2 males and 1 female with a mean age of 60.0±3.05 years. Osteoarthritis was observed in the knee joint in 2 cases and the hip joint in 1 case. Arthroplasty was applied to all the cases. In a mean follow-up period of 72 months no complications or recurrence were observed. Arthroplasty is an effective treatment method in advanced stage osteoarthritis which has developed as a result of ochronosis-related damage.

Keywords: Alkaptonuria, ochronosis, osteoarthritis, knee, hip, arthroplasty.

ÖZET

Homogentisik asit oksidaz enzimi eksikliğine bağlı biriken homogentisik asid eklem kıkırdağında hasarına sonrasında ise yaşlanma ile birlikte osteoartrit gelişimine neden olur. Bu yazıda, okronozis ile ilişkili ileri evre osteoartrit tedavisinde uygulanan artroplastinin sonuçları değerlendirildi. Çalışmaya okronozis ile ilişkili ileri evre osteoartrit geliştirilen ve 2000-2015 yılları arasında artroplasti uygulanan 3 hastanın 6 dizi dahil edildi. Hastaların 2'si erkek ve 1'i kadındı, yaş ortalaması ise 60.0 ± 3.05 yıldı. Osteoartrit 2 olguda diz ekleminde görülürken, 1 olguda kalça ekleminde gözlendi. Tüm olgulara artroplasti uygulandı. 72 aylık ortalama takip süresi içinde herhangi bir komplikasyon veya nüks gözlenmedi. Artroplasti okronozisin neden olduğu hasarla ilişkili gelişen ileri evre osteoartritte etkili bir tedavi yöntemidir.

Anahtar sözcükler: Alkaptonüri, okronozis, osteoartrit, diz, kalça, artroplasti.

INTRODUCTION

Alkaptonuria is a rare metabolic disease with autosomal recessive transfer caused by congenital homogentisic acid (HGA) oxidase enzyme deficiency.[1, 2] HGA which accumulates in the body associated with HGA oxidase enzyme deficiency, accumulates in cartilage, tendons, ligaments, skin, sclera, renal tubular epithelial cells, pancreas islets and in the walls of some arteries[3, 4]. Accumulation in the joint cartilage, together with the ageing process, causes progressive tissue damage and results in osteoarthritis. A blue-black colour change in tissue and the urine together with degenerative arthritis is known as ochronosis[1]. The frequency has been reported as 1/1,000,000 with this rate reaching 1/19,000 in countries such as Slovakia and San Domingo[5].The spine and knee and hip joints are often involved[2]. The first complaint related to the joint is chronic back pain and restricted movement.

Although no medical treatment has been proven to be effective for this disease, there are case reports in literature of surgical treatment on the affected joints. In this paper, 3 cases are presented where arthroplasty was applied because of bilateral knee and hip involvement.

CASE REPORT

In this study an evaluation was made of 6 joints of 3 cases with sufficient follow-up after the application of arthroplasty because of ochronosis-related advanced stage osteoarthritis between January 2000 and January 2015. Evaluation was made in respect of age, gender, affected joint, treatment applied and the outcomes. Informed consent was obtained from all the patients.

The cases were 2 males and 1 female with a mean age of 60.0±3.05 years. Osteoarthritis was observed in the knee joints in 2 cases and the hip joints in 1 case. A 60-year old female and a 62-year old male were siblings (Figure 1). Bilateral joint arthroplasty was applied to all the cases. In a mean follow-up period of 72 months no complications or recurrence were observed. The characteristics of the cases are presented in Table 1. There were intraoperative findings of blue-black colour change in the joint, which was the differentiating feature from primary osteoarthritis (Figure2).

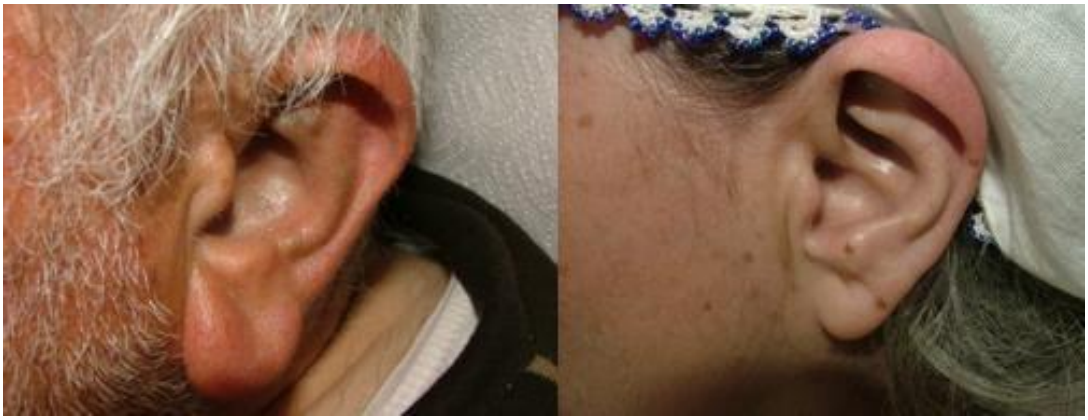

Figure 1. Pigmentations at ears of a 60-year old female and a 62-year old male were siblings.

Table 1. Characteristics of the cases.

	Age (Year)	Gender	Joint	Follow-up period (Months)
Case 1	56	Male	Bilateral knee	72
Case 2	60	Female	Bilateral hip	72
Case 3	62	Male	Bilateral knee	72

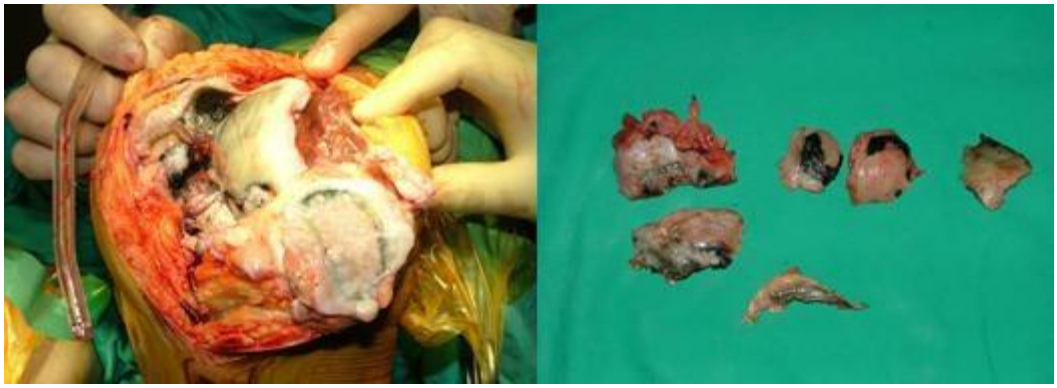

Figure 2. Intraoperative findings of blue-black colour change in the joint.

DISCUSSION

Alkaptonuria was first described in 1584 by Scribonius as a black colour in the urine of children, then in 1866, pigments were seen in microscopic examination by Wirchow and the disease was named ochronosis [2, 6]. At the end of the 1900s, alkaptonuria was determined in the locus of gene 3q21-23 [7]. Alkaptonuric cases are generally asymptomatic in childhood and adolescence. The first finding of the disease is a change in the colour of urine[1]. When there are no ocular or skin findings, this condition can be overlooked. Diagnosis is generally made with the triad of black coloured urine, degenerative arthritis and ochronotic pigmentation[8]

Ochronotic pigment can accumulate in all connective tissue, primarily in cartilage tissue[1].Ochronotic arthropathy is seen mostly at the age of 40-50 years and in joints exposed to weight-bearing[9]. As the first area of involvement is the spine, the first complaints of patients are pain and restricted movement. Apart from the spine, the knee is the most frequently involved area followed by the hip, shoulder, sacroiliac joint and symphysis pubis[1].The cases presented here were aged over 55 years and the involved joints were the knee and the hip. It is extremely rare for the disease to be seen in family members[10]. In the current series, a female aged 60 years and a male aged 62 years were siblings.

Apart from the colour changes associated with joint involvement, the features are similar to primary osteoarthritis[11]. In the current cases, the intraoperative observation of the blue-black colour change in the joint tissues was highly typical of ochronosis.

Although there is no treatment method with proven efficacy, various treatments have been attempted but as the enzyme defect could not be repaired, there has been no chance of recovery in ochronotic arthropathy[12]. Arthropathy is generally treated conservatively but advanced cases may be treated with synovectomy, arthroscopic debridement and arthroplasty[5,13]. Publications in literature about the surgical treatment of affected joints are in the form of case reports. Spencer et al.[14] reported replacements applied to 11 joints of 3 cases. The current series differed from literature in that arthroplasty was applied to 6 joints as bilateral knee and hip joints in 3 cases, 2 of whom were siblings.

In conclusion, ochronosis degenerative joint disease must be kept in mind in differential diagnosis. For patients to lead a normal life, total joint arthroplasty is an effective treatment method in advanced stage osteoarthritis which develops especially in the weight-bearing hip and knee joints.

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