TJFM&PC Turkish Journal of Family Medicine and Primary Care

Case Report / Olgu Sunumu

Genetic Inheritance of Developmental Dysplasia of the Hip: Case Report

Gelişimsel Kalça Displazisi Genetik Geçişi Üzerine: Olgu Sunumu

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ÖZET

Gelişimsel kalça displazisi, kalçayı oluşturan yapıların çeşitli nedenlerle yapısal bozulma gösterdiği dinamik bir hastalıktır. Prenatal, natal veya postnatal gelişebilmektedir. Genetik geçişi önemlidir. Özellikle birinci basamakta hastalığın tespiti gerekmektedir. Bu çalışmada, gelişimsel kalça displazisi tanılı anne ve kız hasta sunulmuştur. Hastalığın özellikle genetik geçişi üzerinde durulmuştur.

Anahtar kelimeler: Gelişimsel displazi, kalça, genetik geçiş

ABSTRACT

Developmental dysplasia of the hip (DDH) is a disease in which the dynamics of the hip structures deteriorates due to various reasons. It may develop in the prenatal, natal, or postnatal period. Genetic inheritance is an important risk factor. The diagnosis of the disease is important, especially in the primary care. Herein, we present a mother and a daughter with DDH to emphasize the genetic inheritance of the disease.

Keywords: Developmental dysplasia, hip, genetic inheritance

Received / Geliş tarihi: 07.01.2017, **Accepted / Kabul tarihi:** 21.02.2017
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Öztürk O. Genetic Inheritance of Developmental Dysplasia of the Hip: Case Report.TJFMPC, 2017;11(2): 136-139.
DOI: 10.21763/tjfmpc.318062

INTRODUCTION

Developmental dysplasia of the hip (DDH), also known as congenital hip dislocation or congenital hip dysplasia,[1,2] is a group of pathologies that includes various hip problems ranging from simple hip instability, in which the primary pathology is capsular laxity, to the complete dislocation of the femoral head from the acetabulum. In all newborn, general incidence is accepted to be 0.5 to 1.5%.[3] It is a debilitating disease since it affects the quality of life adversely by disrupting both physical and psychological health. High frequency in girls, family story, and rise in the incidence of identical twins show the presence of genetic factors. In this study, we present a mother and her daughter diagnosed with DDH to emphasize the genetic inheritance of the disease.

CASE REPORT

A 28-year-old female patient and her 18 months old daughter admitted to the family health care center in December 2015 for a scheduled control. They both had DDH diagnosis. The mother has not received any treatment for DDH. Hip dislocation is seen on the pelvic radiographs of the mother (Figure 1). There was not any trauma, suspicious drug use or prior illness during her pregnancy to her only child. She had given birth to her daughter in the hospital uneventfully. Her physical examination was normal, except walking with a limb due to DDH. She was a second-degree relative of her husband.

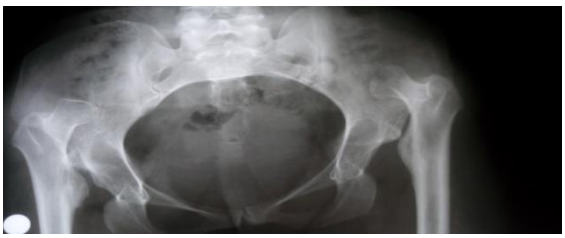

Figure 1. Pelvic radiograph of the mother

The medical history of the baby concerning prenatal, natal and postnatal period was normal excluding DDH. The patient's family physician suspected DDH due to unequal limb lengths in the baby and the positive Barlow and Ortolani examination findings of DDH. Ultrasonography according to Graf method showed both femur heads inside the acetabulum and a blunting of the acetabular bone more obviously in the right side. The alpha angle was 52º and the beta angle was 60º on the right hip. And the alpha angle was 53º and the beta angle was 58º degrees in the left hip. These angle values were compatible with bilateral type 2A hip deformity. After three months follow-up the right hip was consistent with type 1A (normal) hip and left hip with type 2A hip deformity. Pelvic radiograph of the baby showed hip dislocation (Figure 2). The height and weight of both patients were between 25-50 percentiles. No further abnormalities on physical examination were detected. Simple positioning and Pavlik harness treatment were recommended for the baby. Treatment continued at the time of the preparation of this article.

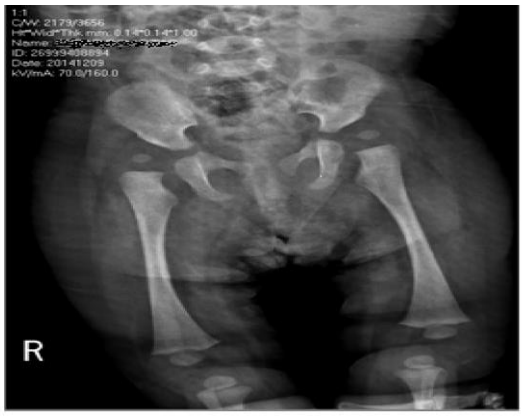

Figure 2. Pelvic radiographs of the baby

DISCUSSION

DDH is a progressive disease which affects hip structures during prenatal, natal or postnatal periods. In a study, 589 patients and their families were examined and the risk of developing DDH due to inheritance was asserted. According to this study, a child from a family without DDH had a lower risk (6%), than one the parents with DDH (12%).[4] Another study showed an increase of a ratio up to 36%.[5] DDH is observed 4-6 times more in girls than boys. While it is three times more frequent on the left hip due to intrauterine position, a bilateral involvement is observed more common if the right side is effected.[6,7] Almost 60% of the children with DDH are first-born child.[8] Our patient is also the first-born child of the family. Being an offspring of a consanguineous marriage, the mother suffering of DDH, female gender, and left hip involvement are in conformity with the literature.

Regarding genetic influence, rather than a specific chromosomal structure, discussing polygenetic predisposition is more accurate.[8] Some associations were detected between DDH and single nucleotide polymorphisms (SNPs) in GDF5, TBX4, and ASPN genes in previous case-control studies in Chinese Han population.[9,10] An association study based on the linkage scan was executed and an SNP in pregnancy associated plasma protein-A2 gene (PAPPA2), rs726252, was found to be associated with DDH.[11,12] Again, it has

also been found in China that HOXB9 is associated with this disease.[13]
Apart from that; many physiological, genetic and mechanical reasons are also known to be involved in etiology. For example, hormonal mediators such as estrogen and relaxin, which have important effects on the mother towards the end of pregnancy, cause a physiological ligament laxity in the newborn. It is accepted that there's a close relationship between breech posture and DDH. In a normal population breech posture incidence is reported to be 3%, wheras in newborns with DDH this incidence rises to 16%.[5] Congenital calcaneo-valgus foot deformity and swaddling of infants are the other major reasons for this entity.[14] Our patient had a normal and spontaneous vaginal delivery; and no additional anomalies were observed. Genetic tests had not been performed.

Clinical examination of a newborn is the primary method for DDH detection. DDH detection are made with Barlow and Ortolani test, where abduction ranges of motion of the hips are evaluated. The Graf method, a radiological imaging method, the alpha and beta angles are determined. The alpha angle is the angle between the iliac bone edge and acetabular roof (normal value > 60 degrees); and the beta angle is the angle between the iliac bone and labrum axis (normal value > 55 degrees[8]). In our patient the alpha angle was increased and the beta angle was decreased.

Closed reduction and body cast can be applied successfully under general anesthesia in children over six months of age. After Pavlik harness application, the patients are followed up with direct radiography every three to four months until skeletal development is completed.[15] After walking age, complicated surgical procedures are required and the rate of success decreases.[3] It is important to note that untreated DDH, particularly when unilateral, may cause asymmetrical joint loading manifesting as leg length inequality, gait abnormalities, and biomechanical effects such as muscle weakness, abnormal joint movement, and knee and foot disorders.[16] It is known that the average age at the time of operation is 35 months and the average follow-up period is 72 months in our region. The family history is 20% positive and swaddling rate is 89%. Therefore, DDH is still a critical condition.[17]

CONCLUSION

Early diagnosis and treatment of DDH, which appears frequently, genetically inherited, disrupting health physically and psychologically, is important. Examination of all newborn for DDH during primary care is necessary. Families should be informed about appropriate baby care and the risks of swaddling. Since treatment of the disease in the early stages prevents complicated surgeries in late stages, early diagnosis will also result in financial advantages.

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