

Gecikmiş Bir Rehabilitasyon Olgusu: Rizomelik Kondrodisplazi Punktata Tip 3

A Delayed Rehabilitation Case: Rhizomelic Chondrodysplasia Punctata Type 3

Yasemin Yumuşakhuyulu, Afitap İçağasioğlu, Erkan Mesci, Ali İhsan Sungur

İstanbul Medeniyet Üniversitesi Göztepe Eğitim ve Araştırma Hastanesi, Fizik Tedavi ve Rehabilitasyon Kliniği, İstanbul, Türkiye

ÖZET

Rizozomal kondrodisplazi punktata (RKDP) lipid biyosentezi için gerekli olan peroksizomal genlerdeki mutasyonlar sonucu gelişen bir hastalıktır. Etkilenen hastalarda proksimal olarak kısa ekstremiteler, epifizyal punktat kalsifikasyon, bilateral katarakt, büyüme ve gelişme geriliği karakteristiktir. Hastaların büyük çoğunluğu RKDP tip 1, %5 civarı ise tip 2 veya 3 tür. Bu makalede RKDP tip 3 tanılı, rehabilitasyonu ihmal edilmiş 9 yaşındaki erkek çocuğu sunduk. Makalemizde, yapılan küçük bir rehabilitatif girişimin kişinin fonksiyonelliğine çok fazla katkıda bulunduğunu göstererek rehabilitasyonun önemini vurgulamak istedik.

Anahtar Kelimeler: rizomelik kondrodisplazi punktata tip 3, rehabilitasyon

ABSTRACT

Rhizomelic chondrodysplasia punctata (RCDP) is a disorder resulting from mutations in peroxisomal genes essential for lipid biosynthesis. Affected patients have characteristic features like proximally shortened limbs, epiphyseal punctuate calcification, bilateral cataracts, growth and developmental delays. The majority of patients have RCDP type 1, around 5% have RCDP type 2 or 3. In this paper we report a case of 9 years old male child diagnosed as RCDP type 3 whose rehabilitation was ignored. We would like to emphasize the importance of rehabilitation which even a slight intervention of rehabilitation can contribute a lot to his functionality.

Keywords: Rhizomelic condrodyplasia type 3, rehabilitation

İletişim / Correspondence:

Dr. Yasemin Yumuşakhuyulu

İstanbul Medeniyet Üniversitesi Göztepe Eğitim ve Araştırma Hastanesi, Fizik Tedavi ve Rehabilitasyon Kliniği, İstanbul, Türkiye

E-mail: yassure@yahoo.com

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INTRODUCTION

Rhizomelic chondrodysplasia punctata (RCDP) is a peroxisomal metabolism disorder, resulting from a deficiency of a specialized class of membrane phospholipids, plasmalogens (1-2). It may be inherited as X-linked dominant, X-linked recessive and autosomal recessive (3). The disease is characterized by a typical facial appearance, congenital cataracts, proximal shortening of the limbs, punctate calcifications of the epiphyses, arthrogenic contractures, growth failure, psychomotor retardation and near absence of developmental milestones (1). Although classic RCDP is commonly considered lethal, with 60% of deaths occurring by age 1 year, a few survive beyond 10 years (4). Recent reports have documented an increased number of children who are surviving many years beyond the expected age (5). The rehabilitations of these children also gain importance together with the extending life span. But the rehabilitation of the RCDP patients is neglected by the reason of short life expectancy and the intensity of internal problems. So we report a 9 years old male child diagnosed as RCDP whose rehabilitation is neglected.

CASE

The authors report the case of a 9 years old child presenting with characteristic facies, rhizomelic micromelia and anthropometric measures below the expected indexes for his age. Skeletal radiographies showed humeral and femoral shortening and punctuate calcifications on shoulder, hip and knee joints. Because of internal problems his rehabilitation need was uncared. The informed consent was obtained from the parents of the affected child.

The baby was born at 38 weeks with the cesarean section as the second living from the third pregnancy of 25 years old mother. He had been stained with meconium when he was born. When he was born, his length was 42 cm (3-50% percentiles), his weight was 2300 gr (3-50% percentiles) and his head circumference was 30.5 cm (3-50% percentiles). He has a one healthy sibling 43n deve other sibling was dead. His parents are first-degree relatives. The mother was under routine prenatal follow-up during pregnancy. There was no exposure of teratogen or drug or

autoimmune disease in the mother during pregnancy. The mother was also negative for diabetes, hypertension, urinary tract infection or early membrane rupture during pregnancy. 46XY karyotype came out in the genetic analysis that had been done by the reason of atypical facial appearance and the punctate calcifications in the direct graphies. The gene analysis came out RCDP type 3 alkyl-dihydroxyacetone phosphate (DHAP) synthase deficiency.

On his physical examination his height was 92 cm and his weight was 9760 gr. He had bilateral cataract surgery and bilateral rales were heard in the lungs. The patient could be fed orally with mashed soft juicy food. He couldn't speak. The other systemic examination findings were normal.

On locomotor system examination he could carry his head. According to modified Ashwoth scoring, there was general joint spasticity at the level of 2 – 3. The ranges of joint motion were limited in both shoulders, elbows, hips and knees. There was no sitting balance (Figure 1). Gross Motor Function Measure [GMFM-88] score was 29.

We applied botulinum toxin-A to his hip adductors, hamstrings and gastrocnemius muscles and instruct the mother range of motion and stretching exercises. In addition, the patient was supplied with a sitting chair and orthoses. His spasticity regressed 1 – 2 degrees within the first week control. Within the first month control we saw 5 – 10 degrees of increase in both hip and knee joints motions. A convenience was provided in toilet cleaning and he began to sit tightly and more comfortably (Figure 2). The family was satisfied with the treatment.

DISCUSSION

Chondrodysplasia punctata (CDP) is associated with a number of disorders, including inborn errors of peroxisomal and cholesterol pathways, embryopathy and chromosomal abnormalities (3,6-8). It includes peroxisome biogenesis disorders (Zellweger syndrome, neonatal adrenoleukodystrophy, infantile Refsum disease and RCDP Type1), maternal conditions and teratogen exposure. CDP has four main types, the autosomal dominant, autosomal recessive (rhizomelic type), the X-linked dominant form and

the X-linked recessive form (9). RCDP is a rare autosomal recessive disorder characterized by severe rhizomelic shortening of limb, punctate calcification of cartilage and premature ossification of the epiphyses and metaphyses, coronal clefts in the vertebral bodies, growth failure and severe psychomotor retardation (10). There are three types of RCDP; Type 1 involves mutations in the PEX7 gene (peroxisomal assembly disorder) (9), type 2 and type 3 have a single enzyme defect, affecting plasmalogen synthesis (1,3). RCDP types 2 and 3 are phenotypically similar to RCDP type 1, but result from deficiencies of dihydroxyacetone phosphate acyltransferase and alkyldihydroxyacetone phosphate (ADHAP) synthase, respectively (9).

Diagnosis of the disease is based on clinical findings and confirmed by clinically available biochemical or molecular genetic testing which includes biochemical tests of peroxisomal function like red cell plasmalogen concentration, plasma phytanic acid and very long chain fatty acid estimation (9). The diagnosis of our patient whose cytogenetic analysis was 46-XY had been made as RCDP type 3 having acyl-DHAP synthase deficiency by the Genetic Metabolic laboratory of the University of Amsterdam.

The disorder is considered sublethal, most of the affected fetuses die in utero or in the first few weeks of life. Approximately 60 % of the cases survive the first and 39 % the second year. Only few of them survive beyond this time with severe developmental delays, physical disability and profound mental retardation and death usually occurs in the first decade of life (6,9,11). Our patient who is 9 years and 6 months old has lived a longer period than expected but his anthropometric measurements are far more behind his age. He could carry his head but there was no sitting balance. There was common spasticity and limitation of range of motion in all joints.

The management of RCDP is principally supportive. Genetic counseling is required. Cataract extraction and physiotherapy could help. Regular assessment on follow up needed for monitoring growth and development, seizure control, vision, hearing, contractures and orthopedic complications (12). The orthopedic evaluation and the

rehabilitation of our patient was ignored by the reason of short life expectancy and the intensity of internal problems.

In RCDP cases, spasticity, psychomotor retardation, growth retardation, seizures, thermoregulatory instability, 44n dev difficulty, recurrent otitis media, and pneumonia have been reported (13). These patients often demonstrate upper and lower extremity spasticity in the absence of spinal cord involvement. Because of limited range of motion in multiple joints it is sometimes hard to establish the spasticity (14). Since our patient has common joint spasticity and the limitation in the ranges of joint motion, botulinum toxin type-A, exercises and orthoses have been proposed.

In summary, we report a case of rhizomelic chondrodysplasia punctata type 3 of whose rehabilitation was neglected. Rehabilitation is the name given to all diagnostic and therapeutic procedures which aim to develop maximum physical, social and vocational function in a diseased or injured person. The goal of the rehabilitation is to gain independence in activities of daily living, school or work and social life. This is possible to the extent of the person's impairments (15). The intensity of the internal problems and the short life expectancy of our patients lead to be less cared about their life quality, functionality and musculoskeletal system problems. But the increased number of children who are surviving beyond the expected age make it necessary to pay more attention on their rehabilitation. 44n deven a tiny contribution, may add a lot of convenience. We treated the patient and added much convenience to his life.

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