Journal of
Modern Rehabilitation
July 2017, Volume 11, Number 3

Case Report : Occupational Therapy in Cockayne Syndrome: A Case Report

Marzieh Pashmdarfard[1*]

1. PhD Student, Department of Occupational Therapy, School of Rehabilitation Sciences, Iran University of Medical Sciences, Tehran, Iran.

Use your device to scan and read the article online

Citation: Pashmdarfard M. Occupational Therapy in Cockayne Syndrome: A Case Report. Journal of Modern Rehabilitation. 2017; 11(3):189-193.

Article info:
Received: 19 Jan. 2017
Accepted: 10 Apr. 2017

Keywords:
Cockayne Syndrome, Occupational therapy, Case report

ABSTRACT

Introduction: Cockayne syndrome is a rare and autosomal recessive neurodevelopmental disorder characterized by symptoms such as progressive neurological disorder, photosensitivity, visual disturbances, microcephaly, premature aging, and birdlike nose, mustache, and early asymmetric eyes. The syndrome has three types; I, II, and III.

Results: The patient is an 8-year-old boy with Cockayne syndrome type II whose parents have a grade 3 relationship. The child received Occupational Therapy (OT) intervention since 9 months of age. When he was one year old he could control his head and at the age of 2.5 years he could crawl. At age 3, the child was able to cruise and express some words like *mama, baba, and meow meow*. However, these abilities are gone now and he is only able to barely sit and control his head. Since the child has continuously received occupational therapy since the age of 9 months, and his CS is of the type two (the worst type), it can be argued that the offered child-care services along with all medical treatment, were successful to slow the disease progress and prevent the occurrence of secondary problems.

1. Introduction

Cockayne Syndrome (CS) was first described by an English physician Edward Alfred Cockayne in 1936 and for the second time in 1946 [1]. Following him, Mary M. Dingwall and Catherine A. Neill described two brothers with the symptoms of the disease described by Cockayne, and that is why the other name of this disorder is the Neill-Dingwall Syndrome [1]. CS is a rare autosomal recessive neurodegenerative disorder characterized by symptoms such as progressive neurological disorder, photosensitivity, visual impairment, microcephaly, premature aging (Progeria), birdlike nose, mouse shaped face and asymmetric eyes [2-4]. Neurological disorders and neurodevelopmental disorders are the diagnostic criteria for this disorder, while photosensitivity, loss of hearing, and visual impairment are among other commonly used diagnostic criteria [5].

Any defect and impairment in the function of the internal organs is possible due to a group of disorders (leukodystrophies) that occurs due to the destruction of the brain white matter [5]. Nevertheless, these patients are not prone to infectious diseases or cancers [6] and their symptoms are due to a defect in the DNA repair

*** Corresponding Author:**
Marzieh Pashmdarfard, PhD Student
Address: *Department of Occupational Therapy, School of Rehabilitation Sciences, Iran University of Medical Sciences, Tehran, Iran.*
Tel: *+98 (21) 22228051*
E-mail: *hazal.ot.fard@yahoo.com*

[5]. Today, the disorder is known as premature aging, Progeria, or Hutchinson–Gilford Progeria Syndrome (HGPS) [1]. The incidence of this disorder is as low as 1 in 8000000 live births – denoting a very rare genetic disorder [7]. People born with CS usually live about 13 years, although many cases of this syndrome have been reported that reach the puberty and even the age of twenty years. However, a few affected people have lived up to 40 years, but the severity of the symptoms and type of syndrome is effective in their survival [7]. Cockayne syndrome is of three types; I, II, and III [8].

Cockayne Syndrome type I

This syndrome is known as the classical type. The patients have normal fetal development, and the first signs of this disorder appear at the beginning of the second year of life. Visual and hearing ability of these patients gradually diminish [8]. The central and peripheral nervous system is rapidly deteriorating and continues until death occurs in the first or second decade of life. Brain atrophy is not very severe in these patients [7].

Cockayne Syndrome type II

The symptoms of CS type II are more severe [8]. Nervous system growth after birth rarely continues and these patients often die at the age of seven. This type of CS is known as Cerebro-Oculo-Facio-skeletal Syndrome (COFS) or Pena-Shokeir syndrome type II [8]. Such naming is because of disease effects on brain (brain atrophy), eye (vision loss or cataract), face (loss of facial fat), and skeletal system (osteoporosis) [8]. Patients who develop symptoms soon after birth show more severe brain damage, such as loss of myelination in the brain white matter, massive calcification in the cerebral cortex and basal ganglia [7].

Cockayne Syndrome type III

In these patients, symptoms develop more slowly and the severity of the disease is milder than those of type I and II, so the patients can survive and reach even adolescents and adulthood period [8].

2. Case Report

The patient is a 9-year-old boy whose parents were blood relatives; the patient was the second and last child of the family, and according to genetic studies, the daughter of the family was the carrier of the disease, too. The mother was 27 years old at the time of pregnancy. According to the mother and child's medical records, the baby was completely normal at the time of birth, and no abnormal signs were observed during pregnancy. The baby's head circumference at the time of birth was 32 cm, his height was 52 cm and his weight was 2600 g. After two months, the physicians informed that the child's head size grew slowly so the child started to receive occupational therapy interventions at the age of 9 months. The child gained head control when he was one year old, and at the age of two and a half years he started crawling and kneeling.

He was cruising during the age of 3, and at that age he was able to babble and express some words like mama and baba. But he lost these abilities over time, and at the time of admission, he could barely sit and control his head. When he was 9 years old, his head circumference was 42 cm, his weight was 9 kg, his height was 85 cm, and was only able to say the word mama unclearly. His abnormal photosensitivity was very manifest, with her face showing a degree of sunburn. Her facial fat had somewhat scratched off, with a clear-cut bird's eye view. Microspheres, kyphosis posture and decrease in the range of motion of shoulder movement in the child became more intense and his hearing loss and vision problem was apparent (Figure 1).

Occupational therapy interventions

When the child was 9 years old, a physician diagnosed his disease, Cockayne syndrome type II. Before this age, he was referred to occupational therapy clinic as a child with microcephaly and cerebral palsy. Because of the nature of CS type II and the severity of its symptoms, the child was expected to get crippled much faster than his age and even be expired by now. However, since the child has continuously received occupational therapy services from the age of 9 months, it is possible that the child-care services especially the offered occupational therapy interventions to the child, along with all good medical treatment, were successful in slowing the disease progress. Since occupational therapy uses a top-down approach to deliver services to patients, the same approach was used in intervention process of this child [9].

Because the disease of this child is progressive, we decided to focus on the mobility and play as his main activities in accordance with his age and family requests. After a comprehensive top-down evaluation, among the performance skills we turned to the motor skill components which are more related to the disability of the child in play and mobility areas of activities. According to the Occupational Therapy Practice Framework (OTPF), OT evaluation and interventions can be implemented at three levels, occupational performance level (such as activities

Figure 1. Patient with Cockayne syndrome, patient's Progeria and kyphosis posture

JMR

of daily living, play, leisure and so on), occupational performance skills level (motor skills, process skills, social interaction skills), and occupational performance components (such as muscle tone, ROM, kyphotic posture, memory and so on) [10] (Figure 2). OT intervention has been done three sessions per week and 45 minutes per session. The OT approach which was conducted on this child was preventive approach.

In order to slow down the development of kyphotic posture in this child, trunk exercises focusing on active trunk movements was conducted based on playful activities. To increase the active shoulder range of motion and the head control of the child, active shoulder exercises and above shoulder surfaces were performed based on playful activities. In order to reduce the muscle tone of the lower extremities and prevent the progression of osteoporosis, the child was asked to do the Weight Bearing (WB) activities with a combination of shoulder movements when he was standing on a board or wedge. In order to increase visual attention, the exercises for recognizing the form constancy and figure – ground perception were conducted on the child by creating the maximum contrast in a dark room. We tried to strengthen the child's visual pursuit by using colored lasers and throwing them on the wall. In order to enhance the auditory attention in the child, we tried to provide a variety of hearing stimuli. For example, we called the child and also used different musical instruments in different directions in order to make him react to the sound and

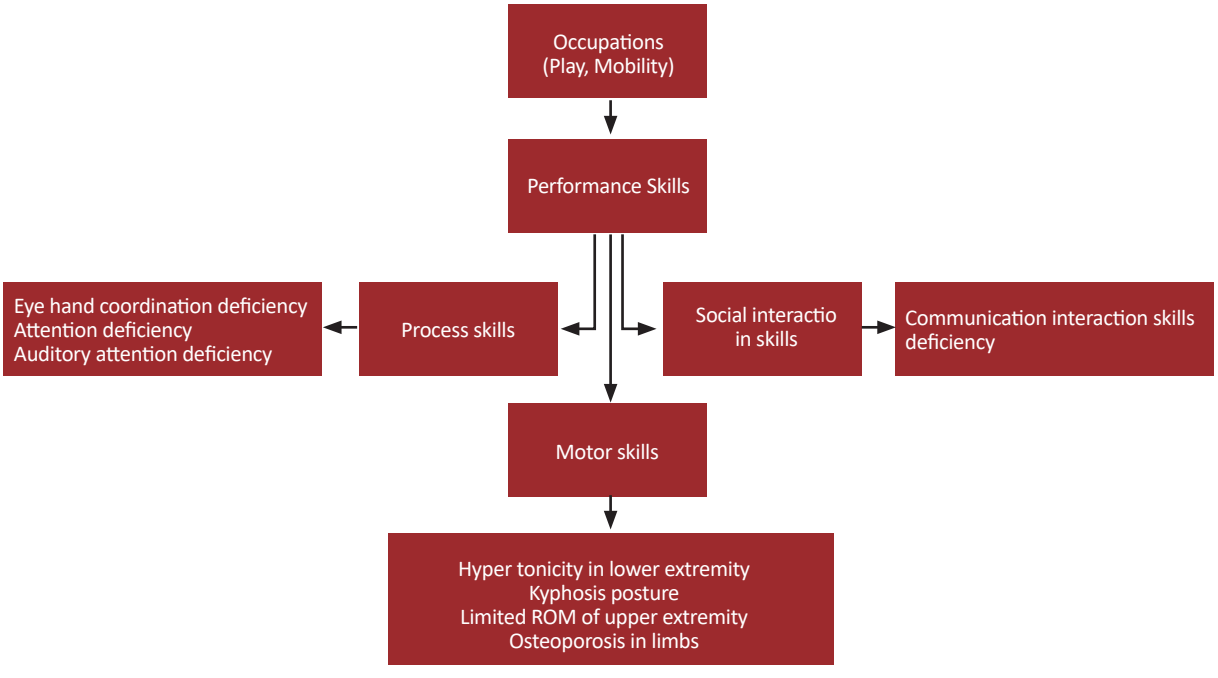

Figure 2. Summary of a comprehensive top-down evaluation of child

JMR

strengthen the child's auditory attention. At each session, we tried to provide the home exercises for child's parents and the advice on the next pregnancies, and informed them that if pregnancy is desired, the genetic tests should be carried out.

3. Results

All the results of this study were based on therapist observation and parents' reports. Berceuse of long duration of occupational therapy intervention (about 8 years) we didn't use any standard assessment scale. Therapist's observation demonstrated that the preventive approach in this child had good results and the parents and physician reports confirmed these results, too. Because of the nature of the disease, the family physician expected the child be expired very soon; however, based on his physician's reports the OT interventions were very useful and beneficial for this child.

4. Discussion

There is no study so far about the rehabilitation intervention on CS and a few studies on this syndrome only discussed medical and genetic symptoms of this disease. Thus we hope that this study as the first study about the rehabilitation intervention on CS be a useful study to introduce this syndrome and some of its useful rehabilitation interventions.

The main purpose of the rehabilitation team, especially occupational therapists, is to achieve maximum independence and improve the quality of life of the referring patients and their caregivers [9]. Therefore familiarity with some rare disorders, as well as the prognosis of the treatment process and interventions, can provide useful information for the occupational therapists and give them best possible treatment services in dealing with their clients. Occupational therapy requires special attention to genetic disorders as well as progressive disorders. In these cases, with the knowledge of the disorder prognosis, treatment and intervention can be better managed. As one of the symptoms in these patients is osteoporosis it is best to avoid over stretching and over exercising. In order to increase the life-expectancy in these patients it is advisable to prevent undesired postures such as kyphosis and scoliosis by prescribing proper splints, assistive devise, and proper treatments. Since the reduction in visual acuity in these individuals is symptomatic, it is better to reduce the degree of visual acuity problems by creating accurate contrast and creating the desired brightness in their living environments, as well as the lack of attention of the patient to the environment [11]. Regarding the possibility of parents' illiteracy about the heredity nature of the disease, their education can be useful to avoid the problem in next pregnancies.

With proper knowledge of the disorder and illness, treatment and interventions will be more feasible and appropriate. CS is a rare genetic disorder that can be diagnosed among occupational therapists' clients by searching symptoms such as delayed developmental and motor developmental stages (due to the nature of the disease). Occupational therapists should pay attention to these symptoms in their patients and then they can choose the best treatment and interventions.

Acknowledgments

This research did not receive any specific grant from funding agencies in the public, commercial, or not-for-profit sectors. The author appreciate the parent of studied child.

Conflict of Interest

The author declared no conflicts of interest.

References

[1] Neill CA, Dingwall MM. A Syndrome resembling progeria: A review of two cases. Archives of Disease in Childhood. 1950; 25(123):213-23. doi: 10.1136/adc.25.123.213

[2] William J, Berger T, Elston D. Andrews' Diseases of the skin: Clinical dermatology. Amsterdam: Elsevier; 2005.

[3] Mallory SB, Krafchik BR, Bender MM, Potocki L, Metry DW. Cockayne Syndrome. Pediatric Dermatology. 2003; 20(6):538–40. doi: 10.1111/j.1525-1470.2003.20619.x

[4] Hoeijmakers JHJ. DNA damage, aging, and cancer. New England Journal of Medicine. 2009; 361(15):1475–85. doi: 10.1056/nejmra0804615

[5] Nance MA, Berry SA. Cockayne syndrome: Review of 140 cases. American Journal of Medical Genetics. 1992; 42(1):68–84. doi: 10.1002/ajmg.1320420115

[6] Bertola DR, Cao H, Albano LMJ, Oliveira DP, Kok F, Marques-Dias MJ, et al. Cockayne syndrome type A: Novel mutations in eight typical patients. Journal of Human Genetics. 2006; 51(8):701–5. doi: 10.1007/s10038-006-0011-7

[7] Javadzadeh M. Cockayne Syndrome. Iranian Journal of Child Neurology. 2014; 8(4):18-19. doi: 10.22037/ijcn.v8i4.7526

[8] Genetics Home Reference. Cockayne Syndrome [Internet]. 2015 [Updated 2015 April 30]. Available from: https://ghr.nlm.nih.gov/condition/cockayne-syndrome

[9] Pashmdarfard M, Amini M, Hassani Mehraban A. Participation of Iranian cerebral palsy children in life areas: A systematic review article. Iranian Journal of Child Neurology. 2017; 11(1):1-12. PMID: 28277550

[10] American Occupational Therapy Association. Occupational Therapy Practice Framework: Domain And Process, 3rd Edition. American Journal of Occupational Therapy. 2014; 68:S1-S48. doi: 10.5014/ajot.2014.682006

[11] Pashmdarfard M, Amini M, Amini M. Rehabilitation of blind people and people with low vision in Iran. Iranian Rehabilitation Journal. 2016; 14(2):77-84. doi: 10.18869/nrip.irj.14.2.77