**MEDICAL REHABILITATION IN PATIENT WITH DOWN SYNDROME**

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**ABSTRACT**: Down syndrome (DS) or trisomy 21 is genetic disorder characterized by physical delay development and intellectual disorder (due to abnormality chromosome 21).1,2 In turn, World Health Organization estimates for DS range between 10 to 11 in 10,000 live births worldwide.1,3 According to the Indonesian Down Syndrome Association (ISDI), data on people with down syndrome in Indonesia up to January 2011 reached around 350,000 cases and constituted 15% of the total cases of world down syndrome. In this case report is about rehabilitation a child with Down syndrome treated at the Medical Rehabilitation Prof. R.D Kandou hospital Manado. In this case, the problem experienced by patients is the problem of developmental delay, hypotonia and speeech.29 Management in physical medicine and rehabilitation for children with down syndrome includes management of cognition, communication, motor, emotional and social development and parenting. The short-term goal has been achieved, but therapy for this patient is still on going with a number of planning therapies to achieve long-term goals.4,10,37

**Key words**: Chromosome, down syndrome, rehabilitation.

**ABSTRAK:** Down sindrom (DS) atau trisomy 21 adalah kelainan genetik yang ditandai dengan keterlambatan perkembangan fisik dan gangguan intelektual ( karena abnormalitas kromosom 21 ).1,2 Organisasi Kesehatan Dunia memperkirakan kisaran DS antara 10 hingga 11 dalam 10.000 kelahiran hidup di seluruh dunia.1,3 Menurut Indonesian Down Syndrome Association (ISDI), Data penderita down syndrome di Indonesia hingga Januari 2011 mencapai sekitar 350.000 kasus dan merupakan 15% dari total kasus down syndrome dunia. Case report ini membahas tentang rehabilitasi anak down syndrome yang dirawat di Rehabilitasi Medik RS Prof. R.D Kandou Manado. Dalam hal ini masalah yang dialami pasien adalah masalah keterlambatan perkembangan, hipotonia dan bicara.29 Penatalaksanaan di bidang kedokteran fisik dan rehabilitasi anak down syndrome meliputi pengelolaan kognisi, komunikasi, motorik, perkembangan emosi dan sosial serta pola asuh. Tujuan jangka pendek telah tercapai, namun terapi untuk pasien ini masih berjalan dengan beberapa terapi perencanaan untuk mencapai tujuan jangka panjang.4,10,37

**Kata kunci:** Down syndrome, kromosom, rehabilitasi.

**INTRODUCTION**

Down syndrome (DS) or trisomy 21 is genetic disorder characterized by physical delay development and intellectual disorder (due to abnormality chromosome 21).1,2 In turn, World Health Organization estimates for DS range between 10 to 11 in 10,000 live births worldwide.1,3 According to the Indonesian Down Syndrome Association (ISDI), data on people with down syndrome in Indonesia up to January 2011 reached around 350,000 cases and constituted 15% of the total cases of world down syndrome.4

At the current time, children with DS are almost always referred for early intervention programs after birth. The most common early intervention services for babies with DS are physical therapy and speech therapy.8 This requires early intervention involving the medical rehabilitation team, including physiotherapy, occupational therapy, prosthetic orthotics, speech therapy, psychology and medical social which are tailored to the problems that are found in patients. The aim of rehabilitation interventions early is to maximize developmental potential, and improve long – term

functional capabilities so as to gain independence.4

**CASE REPORT**

**INITIAL PRESENTATION**

A 8 months old baby girl was referred to outpatient clinic medical rehabilitation Prof. R. D. Kandou Hospital with chief complaint can't sit independent. Patient until now have not been able to sit independent. During sleep the patient can rolling by her self since the age of 5 months but cannot crawling, just creeping up about 1 meters. Patients until now have not been able to smile spontaneously and have not responded to play together. Since more than 1 month ago the patient has been able to look at her parents and look for sources of sound and cannot mumble yet. Until now patients eat porridge filter because she has difficult to feed. Patient loves to drink milk from a bottle but she can’t hold the bottle by herself. The patient is daily cared for by her mother and grandmother. The patient diagnosed with down syndrome since birth and pediatricians do chromosome checks, positive results are found to experience a down syndrome.

The patient had experienced seizure when she

was 3 months. The mother runs a pregnancy check up regularly, controls 9 times at the obstetrician and public health. During pregnancy the patient's mother is healthy. Tetanus toxoid immunization is done twice. The patient was born when her mother was 38 years old with Sectio Caesaria caused by plasenta previa in 36 months pregnancy, with a Birth Weight of 2700 grams and body length of 44 cm. After born, patients not crying right away and treated incubator for 10 days.Patient can’t crawling, sitting, standing, laughing, bubling until now but she can head rising on 3 months o and turning prone – supine and supine – prone on 4 months old.Status immunization was complete. On family history there is the cousin of the patient's mother have same condition.

At physical examination weight 6,7 kg, height 53 cm, head circumference 35,5 cm, normal vital parameter, characteristic facies, gap between toe and second finger. Primitive Reflexes like moro, sucking, ATNR (Assymetrical Tonic Neck reflex), grasping were negative. Development based on milestone special for down syndrome gross motor : holds head steady balanced, rolls over, can’t sit without support, can’t crawling . On fine motor / adaptive : patients have been able to see and follow objects, the patient can graps dangled ring, cannot passes object hand to hand. On communication/language milestone : the patient has reacted and sometimes turns towards the sound, cannot say canonical babbling. On social developmental milestone : sometimes patients can smile if given a stimulus but have not been able to smile spontaneously. Neuromuskular examination was normal.On blood test showed increased free T4, echocardiography atrial septal defect (ASD) and ventricular septal defect (VSD), chromosome analysis 21 chromosome abnormalities due to non-dysjunction (47, XX, +21)

**REHABILITATION COURSE**

Rehabilitation problem of this patient is hypotonia, cannot crawling, sitting inadequate, limitations in playing and communication, the parent worry about the development of their child's growth and social interaction with the community in the future.

Goal therapy for this patient consist of short term goal and long term goal. Short term goal were patient able to sit by her self, good sitting balance, patient can crawling, prevent complication, patient able to communicate with her closed family (parent and brother), patient don’t have feeding problem, parents accept patient’s condition and the long term goal were patient able to walking independently without assisted, improve ADL independent such as development of self-care that is suitable for independence and skill, patient able to communicate and interaction with other people, patients can attend special schools (YPAC).

Program rehabilitation we give to this patient were exercise to improve postural control and stability, muscle strength, massage therapy, exercise to increasing muscle tone, exercise to improve mobility skills, exercise to increasing coordination and balance, exercise to increase ADL and playing activity, oromotor exercise, speech therapy, standing exercise.

In this case, based on the follow up of the patient from the first time patient cannot sitting by her self, after 3 months therapy patient can sit independenly and after 7 months therapy patient can changing position from sleeping to sitting by her self and can standing with help/assisted. After 8 months therapy patient can crawling about 2-3 meters and can standing with help.

**DISCUSSION**

In this case report, a 8 months old baby girl diagnosed with Down Syndrome. Down syndrome

or trisomy 21 is genetic disorder characterized by physical delay development and intellectual disorder (due to abnormality chromosome 21).1,2 In turn, World Health Organization estimates for DS range between 10 to 11 in 10,000 live births worldwide.1,3 According to the Indonesian Down Syndrome Association (ISDI), data on people with down syndrome in Indonesia up to January 2011 reached around 350,000 cases and constituted 15% of the total cases of world down syndrome.4

The diagnosis of Down syndrome in this case is based on history, physical examination, and confirmed by chromosome analysis. In this patient, from anamnesis there was a delay developmental disorder, and the age of the mother during 38 years of pregnancy, statistically, the down syndrome was mostly found in women over the age of 35. In general physical examination we found patient had hypotonia, Mongoloid face, joint laxity, strabismus, epicanthal folds, flat nasal bridge, low set of ear, macroglossia, simian crease in his palm, short fingers, dysplasia of midphalanx of the fifth finger and gap between toe and second finger. This patient had Atrial Septal Defect (ASD) and Ventricular Septal Defect (VSD), according to the characteristics of the down syndrome.10,17,18,19

The patient diagnosed with down syndrome and pediatricians do chromosome checks, positive results are found to experience a down syndrome. The exact diagnosis of chromosomal abnormalities in these patients is established after the results of chromosomal analysis of chromosome 21 abnormalities are dueto non-dysjunction, where there is an excess of one chromosome 21 (trisomy 21).20,21

DS is usually detected by prenatal screening, hypotonia at birth, and clinical features. Hypotonia, which is nearly universal at birth, tends to improve with age. Hypotonia and joint laxity significantly delay the development of gross motor skills and increase the risk for joint dislocations. Global developmental delay is the usual presentation. Musculoskeletal disorders are provoked mainly by hypotonia and its consequent muscular weakness, which is more pronounced during adolescence and in the adult years, since in this period of their lives, these people become less active. The muscle strength of the upper limbs and of lower limbs is 50% less in people with DS when compared to the general population. This muscle strength deficit, besides provoking musculoskeletal alterations, is also responsible for a negative impact on the ability to perform daily life activities. DS has a special developmental milestone. Based on the development milestone, it is divided into gross motors, fine motors, communication lags, social and self help.22,23,24 In this patient from examination of voluntary movement for gross motor function: cannot sit without support, cannot crawling. For fine motor function: patient cannot passes object hand to hand. For communication/language milestone : the patient has reacted and sometimes turns towards the sound and patient cannot say canonical babbling. For Social developmental milestone : patients can smile if given a stimulus but have not been able to smile spontaneously. Assesment of milestones revealed a delayed on all item.

Management in physical medicine and rehabilitation for children with down syndrome includes management of cognition, communication, motor, emotional and social development and parenting. Therefore the physiatrist collaborates with physiotherapy, occupational therapy, orthotic prostheses, speech therapy, psychology and social medicine to manage children with down syndrome depend to the problems experienced. So physiatrist must make a program such as arrange rehabilitation goals and planning and prescribe evaluate and modified program so that all the problems are resolved properly.4,10,28

In this case, the problem experienced by patients is the problem of developmental delay, hypotonia and speeech.29 Physical theraphy focus on motor development is overcoming posture control and hypotonia. The goal of pediatric physiotherapist is to teach children with down syndrome to move their bodies in appropriate ways, and to improve their muscle tone. Working with their muscles and movement will help children reach some of their motor milestones and will prevent them from developing problems, such as bad posture, that can accompany low muscle tone. Pediatric physiotherapist do some type of exercise to improve postural control, muscle strength, exercise to improve muscle tone exercise to improve motor control, exercise to improve mobility skills, and exercise to increasing coordination and balance. Pediatric physiotherapist do massage therapy with the aim of stimulation to increase muscle tone and influence motor skills. Massage therapy involves contact between the skin and the skin including stretching, suppression, sweeping, flexion-extension movements in the extremities. This is evidenced from the research by Linkous & Stutts in 13 patients with syndrome down the age of 1-4 years with massage therapy carried out for 30 minutes 2 times per week for 8 weeks showing an increase in muscle activation and improvement in muscle tone. 4,10,30 In this case, based on the follow up of the patient from the first time patient cannot sitting by her self, after 3 months therapy patient can sit independenly and after 7 months therapy patient can changing position from sleeping to sitting by her self and can standing with help/assisted. After 8 months therapy patient can crawling about 2-3 meters and can standing with help.

Children with syndrome down time experience delays in adaptive skills and independence associated with delays in other development domains, such as cognition and motor (gross and fine motor). Occupational therapist focus on the child’s ability to master skills for independence. These can include : self care skills, fine and gross motor skills, play and leisure skills. In this case, in occupational therapy exercise-ball / gymnastic-ball exercise with the aim of strengthening the neck muscles and trunkus. Karimi et all's study found that sensory stimulation simultaneously and occupational therapy can improve motor abilities. Occupational therapy also adding toys placed on the table part so patient can stimulate to do play exercise.31 And based on the follow up, patients experience a significant increase where before therapy patients cannot smile spontaneously and play activities like children of their age, after therapy for about 8 months, now patients can play activities and interact with family, patient can play toys together, clapping hand and smile while playing.

Although most children with down syndrome learn to speak and will use speech as their primary means of communication, they will understand language and have the desire to communicate well before they are able to speak. Because children with down syndrome often have small mouths and slightly enlarged tongues, they can have trouble speak clearly. A speech therapist will work with an individual to help them to learn to communicate clearly. This can be achieved through talking and oromotor exercise stimulation.19,32 In this patient until now (8 months therapy) still cannot speak or say canonical bubling.

Psychological and social aspects of medicine are important as a support in the management of down syndrome patients, because usually parents in down syndrome patients experience shame, anxiety and inferiority. In the social medical aspect there is a need to evaluate and educate the surrounding environment, and motivate parents and children to join the down syndrome community in Indonesia, for example the Parents Association of Down Syndrome Children (POTADS) and the Indonesian Down Syndrome Association (ISDI). The role of psychology is important to provide motivation to parents to stay motivated in fostering, training and raising children with down syndrome.10,33,34

Also needed is the level of knowledge and insight of parents and education in handling children with down syndrome. Education, among others approach techniques that can stimulate cognitive development, for example: parents move objects slowly in front of the child's eyes so that the child can follow the movements of the object, pronounce the child's name as often as possible and teach the child the name or label of the items he sees, parents can read and sing for children regularly.35,36

Early intervention through medical treatment has likely been a major factor that has helped improve health and quality of life of children with DS. Corrective surgery for heart defects, gastrointestinal irregularities, screening for visual impairment, ear infections, hearing loss, hypothyroidism, and obesity are amongst the early medical treatments individuals with DS can benefit from given the comorbidities associated with their condition.9

In this case, the short-term goal has been achieved, but therapy for this patient is still on going with a number of planning therapies to achieve long-term goals.4,10,37

When these people receive good medical care and stimulation, they have the potential to be fully included socially. In this case, currently the prognosis for ad vitam and ad sanationam is bonam due to overall condition of the patient, ie good nutrition, absent of respiratory problems and the presence of adequate support from the family. And prognosis for ad functional is bonam for walking and communication are dubia ad bonam.4,10,38

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