Rehabilitation in Down Syndrome Patient with Malnutrition, Sensory Processing Disorder, Obstructive Sleep Apnea: A Case Report

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Abstract: Down syndrome children will experience delays in development. However, will achieve same milestones as other children on their own timetable, has same motor development sequence but it takes two times compared to healthy children. Management for comorbidities accompanied Down syndrome child is needed. Case: A 4 year 1 month girl with chief complaint could not walk. Patient was diagnosed with Down syndrome, malnutrition, sensory processing disorder, obstructive sleep apnea. She got neurodevelopmental therapy, lower extremities strengthening exercise, sensory integration therapy, consulted to nutritionist and ENT department. She was suggested to use NGT and had an adenoidectomy but the mother refused. After 3 months, she could stand without support but still could not walk. Discussion: Down syndrome causes delay in development, comorbidities make it worst. Patient had delays compared to Down syndrome developmental graphic. There were improvement in gross motor but malnutrition, adenoid hypertrophy, obstructive sleep apnea interfere the management. Conclusion: Down syndrome usually has comorbidities. Down syndrome causes delay in development and comorbidities make it worst. It is important to identify the comorbidities and make holistic approaches.

1 INTRODUCTION

Down syndrome was first described by an English physician John Langdon Down in 1866, but its association with chromosome 21 was established almost 100 years later by Dr. Jerome Lejeune in Paris.

It is the presence of all or part of the third copy of chromosome 21. It is coupled with mental retardation, congenital heart defects, gastrointestinal anomalies, weak neuromuscular tone, dysmorphic features of the head, neck and airways, audiovestibular and visual impairment, characteristic facial and physical features, hematopoietic disorders and a higher incidence of other medical disorders (Kazemi, Salehi and Kheirollahi, 2016).

The Centre for Disease Control and Prevention (CDC) estimates that each year about 6000 babies are born with Down syndrome (an average of 1 in 691 infants born in the United States), without any predilection of race or socioeconomic class (CDC, 2011). In Indonesia, according to Riskesdas, the 2013 prevalence of Down syndrome in children 24-59 months old is 0.13%, and on 2018 is increasing to 0.21% (Wardah, 2019).

The likelihood for having a baby with Down syndrome increases with advanced maternal age and the highest odd is when the mother reaches 40 years old (Ivan and Cromwell, 2014).

Some of the traits common to babies with Down syndrome include low muscle tone, a flat facial profile, short neck, bulging tongue, a small nose, an upward slant to the eyes, a single deep crease across the centre of the palm, an excessive ability to extend the joints, small skin folds on the inner corner of the eyes, excessive space between large and second toe. But the features associated with Down syndrome can be found in babies without Down syndrome, it is needed to order a test called a chromosomal karyotype to confirm diagnosis. Using a sample of blood, this test analyses the child's chromosomes. If

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there's an extra chromosome 21 in all or some cells, the diagnosis is Down syndrome.

Short stature is a cardinal feature of Down syndrome. The growth retardation of children with Down syndrome commences prenatal. After birth growth velocity is most reduced between 6 months and 3 years of age (Myrelid *et al.*, 2002). Statural growth is a well-known indicator of health during childhood. As growth and final height differ markedly between children with Down syndrome and healthy children, standard growth charts should not be used for children with Down syndrome. Concomitant diseases, such as laryngopharyngeal reflux, dysphagia, and sensory processing disorder may influence growth in this patient.

Development is a continuous process that begins at conception and proceeds stage by stage in an orderly sequence. There are specific milestones in each of the four areas of development (gross and fine motor abilities, language skills, social development and self-help skills) that serve as prerequisites for the stages that follow. Most children are expected to achieve each milestone at a designated time, which can be calculated in terms of weeks, months or years.

Milestones	Down syndrome		Normal children	
	Average (mo)	Range(mo)	Average(mo)	Range(mo)
Smiling	2	1.5 - 3	1	0.5-3
Rolling over	6	2 - 12	5	2 – 10
Sitting	9	6 - 18	7	5 - 9
Crawling	11	7 - 21	8	6 - 11
Creeping	13	8 - 25	10	7 - 13
Standing	10	10 - 32	11	8 - 16
Walking	20	12 - 45	13	8 - 18
Talking, words	14	9 - 30	10	6 - 14
Talking, sentences	24	18 - 46	21	14 - 32

Figure 1: Milestones of Down syndrome and normal children.

Because of specific challenges associated with Down syndrome, children will likely experience delays in certain areas of development. However, they will achieve all of the same milestones as other children, just on their own timetable. See Figure 1. Children with Down syndrome followed the same motor development sequence and generally took the twice the time for reaching gross motor developmental milestones compared to healthy equals (Kim *et al.*, 2017). In monitoring the development of a child with Down syndrome, it is more useful to look at the sequence of milestones achieved, rather than the age at which the milestone is reached.

Children with Down syndrome have a different growth pattern to that of the general population, their average height is shorter, their head circumference is smaller and their growth rate is slower between ages of 3 – 36 months.

It is essential that growth in children with Down syndrome is carefully monitored. Height and weight should be plotted using the growth charts specifically designed for children with Down syndrome from birth to 18 years.

Infants and children with Down syndrome can have feeding and drinking difficulties. A smaller oral cavity and low muscle tone in the facial muscles can be contributing factors. In addition, the tongue may appear larger due to a high arched palate, a smaller oral cavity and reduced muscle tone in the tongue. Teeth tend to appear at a later stage. Many children are mouth breathers due to smaller nasal passages, and may have difficulties coordinating sucking, swallowing and breathing whilst feeding. All of these factors can impact on how a child develops efficient oral and feeding skills.

Feeding problems in early childhood are shown to have a negative impact on development and can be a source of caregiver stress. Although these kinds of problems occur frequently across the population, their incidence is much higher in children with developmental disabilities than it is in typically developing children. It is estimated that 57 percent of infants with Down syndrome have feeding difficulties during the neonatal period (van Dijk and Lipke-Steenbeek, 2018).

Children with Down syndrome often have anatomical and physiological anomalies, such as a smaller mouth cavity, a smaller upper jaw, dental anomalies, weaker lip tension, and stronger tongue tension. This leads to oral motor problems in roughly four out of five children with Down syndrome. It has also been shown that children with Down syndrome chew less effectively and that tongue protrusion is frequent, which can lead to food being expelled from the mouth and evoke a pharyngeal reflex. Spoon-feeding is also more difficult, as the sucking response remains present for longer in children with Down syndrome, making it hard for them to take an active bite. There is a delay in the development of oral motor skills needed to eat solid food.

In addition, it has been shown that 45% of children with Down syndrome show selectivity by texture and less self-feeding. In addition, children with Down syndrome display behavioral problems during feeding more often than typically developing children.

Children who have increased oral sensitivity often have difficulty accepting new tastes and textures. Some children with Down syndrome may require support for feeding difficulties, poor weight gain, and oral sensitivity.

2 METHODS

A 4-year-1-month-old girl diagnosed at neonatal period with Down syndrome (47,XX,+21), with no other relevant personal or family history, was brought to the physical medicine and rehabilitation department with chief complaint unable to walk.

The child was unable to walk but able to sit without support. For the fine motor, she was able to hold toys but was not able to reach. She still could not talk, only babbling. She could understand simple instruction, such as to shake hand or to do high five. The instruction must be given in loud voice because she had hearing problem diagnosed as otitis media with bilateral effusion. Right now the otitis media is already on therapy. She was able to smile to her mother.

The mother also reported that the child had feeding difficulty, refusing to eat textured food. She still ate porridge. The mother said that patient could only eat a little amount of food. For the habit of feeding, she was given food by her mother 3 times per day and always in upright position. She could only ate 3 spoons of porridge and 3 x 120 ml of formula milk but sometimes she did not drink all the milk. The mother said that if the food given was too much the patient would vomit.

When she was sleeping, sometimes she suddenly awakened and gasped then continue to sleep. The oxygen saturation measurement for 4 days during night sleep sometimes reach 90%. She slept for around 10 hours a night but disturbed by the waking up episodes 3-4 times. In the morning she got up cranky and crying when her mother woke her up. At daytime, she looked sleepy and sometimes fell asleep during playtime. She also got tired easily and then got cranky. She took a nap for around 2 to 3 hours. Her snoring got worse if she had upper respiratory infection, and she woke more frequent at night.

On physical examination, the girl appeared ill, was inactive and pale, and presented sparse, thin hair. She seemed to have slow reaction time and trouble in paying attention.

For the neuromuscular examination, there were no upper motor neuron sign. The muscle seemed hypotrophy and felt hypotonus. There were hyperlaxity on both extremities but there were no deformity. Heart examination showed normal result. She produced harsh sounds when she breathed.

Her height and weight was <5th centile, as measured for age and sex according to Down syndrome growth charts (Figure 2).

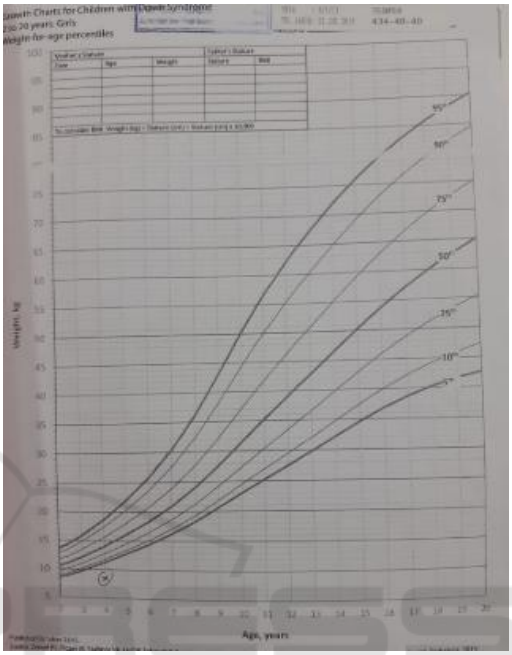

Figure 2: Down syndrome growth chart.

No other changes were observed on systemic examination, except for the physical features of Down syndrome (slanting eyes, epicanthic folds, high-arched palate and protruding tongue). Echocardiography result was normal. The thorax radiology was on normal appearance.

Patient was also diagnosed for sensory processing disorder after filling the sensory profile questionnaire. She had tactile disorder on the registration and sensitivity quadrant. Right now she was on sensory integration program.

She also had adenoid hypertrophy. From the FEES on August 22nd, 2019 there were adenoid hypertrophy 90% and oral phase mechanical dysphagia. From psychological examination, it was obtained that she had severe intellectual disability, Intellectual Quotient 25-35.

Patient already got neurodevelopmental therapy for 3 months. She used bicycle and walking by pushing chair to strengthen her lower extremities. The mother was told to do neurodevelopmental therapy every day; 3 times per day for approximately 20 minutes and strengthening exercise every day, 3 times per day for 20 minutes.

3 RESULTS

Using a sample of blood, this test analyses the child's chromosomes. If there's an extra chromosome 21 in all or some cells, the diagnosis is Down syndrome. In this case, the patient has the traits appearance and confirmed by the chromosomes evaluation (Figure 3).

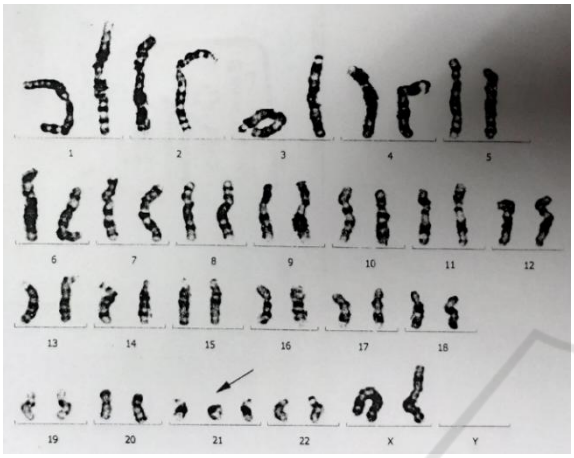

Figure 3: Chromosomal analysis.

In this patient, for the gross motor function, she still cannot walk by herself by the age of four but she already can walk by holding to the wall. For children with Down syndrome the difficulty in walking could be due to hyperlaxity and hypotonicity. For the delayed walking ability compared with Down syndrome milestone can be caused by comorbid condition such as malnutrition. Although this patient had severe cognitive impairment but the impairment does not significantly correlate with the late of gross motor function. Seong et al, 2017, could not find a statistically significant correlation between the achievement of motor milestones and cognitive functions (Kim *et al.*, 2017). Right now, she still could not walk by herself but could stand up and walk by holding to something.

After receiving 3 months of therapy that include strengthening therapy and neurodevelopmental therapy, the progress obtained in this patient was poor. Within three months, she only could stand by holding to the wall.

The patient also had sensory processing disorder with hypersensitivity in tactile and vestibular in registration quadrant. From literature, Down syndrome patient had significant challenges in the categories of low energy, weak, under-responsive, seeks sensation, auditory processing, and tactile sensitivity (Bruni *et al.*, 2010). This patient had intolerance to several food texture, picky eater, only ate soft textured food such as porridge. She also did not like being hugged by other people except her mother, she could not tolerate her own hair touching her face. This sensory processing disorder worsened the feeding problem.

In this patient, she snores when sleeping, easily sleepy at daytime and always cranky when gets up in the morning. After FEES examination, it was found that there is 90% adenoid hypertrophy. These findings will contribute to sleep breathing disorder symptoms. Clinical manifestation of daytime sleepiness will interferes her learning ability as will reduce attention, memory and mood to do activities, therefore stimulation time that can be done daytime reduced. Consultation to ENT-HN specialist is needed to know if there's indication for tonsiloadenoidectomy surgery in this patient as it can be a focus infection for frequent upper respiratory tract infection.

Positioning is an important thing to do for this patient, the mother should keep in mind to keep her upright when feeds her and one hour after feeding to prevent reflux that will worsen adenoid hypertrophy. Sleeping position also should be evaluated, since she also has relative macroglossia and sleeping in supine position will be a risk for upper airway obstruction during sleep, so sleeping in prone or side lying will be better for her.

4 DISCUSSION

Children with Down syndrome has delays in all area of development. For gross motor development, children with Down syndrome will have the same sequence of development compared with other healthy equals but will take a longer time to achieve it. We use Down syndrome developmental chart for the patient. In this study we found that she has delays according to Down syndrome chart.

The possible factors that could cause this problem are the malnutrition state, the adenoid hypertrophy, the obstructive sleep apnea, and also could be cause by the low education of the mother.

This patient also had sensory processing disorder in tactile and vestibular registration and sensitivity. Sensory processing disorder, especially in tactile area also affected the malnutrition. It is because in patient with tactile registration sensory processing disorder, the child will feel very uncomfortable if there were food in her mouth. This condition worsen the intake and as the result, it will worsen the malnutrition.

The patient already got neurodevelopmental therapy and strengthening exercise for the lower extremities. After three months, there were only small progress, the child was able to stand with support. Neurodevelopmental therapy is a manual and rehabilitative approach to optimize the movement system based upon the scientific principles of developmental kinesiology. It brings the supporting joints and segments into a functionally aligned position. If one muscle is dysfunctional (weak), the entire stabilizing function is disturbed and the quality of the movement is compromised. Rehabilitation programs in this patient after three months show poor progress.

We need to evaluate what causes the poor progress. The first possible cause could be the mother was not giving adequate dose of exercise. It was told that the child should do the neurodevelopmental therapy three times per day for 20 minutes and strengthening exercise three times per day for 20 minutes. The mother said the child could not do the exercise that being told because the child was being uneasy and cranky after a short time of exercise.

Other possibilities that could cause the slow progress in this patient was due to the adenoid hypertrophy. Patient already underwent FEES examination and from that examination we found 90% of occlusion. It could cause inadequate oxygenation into the patient's organ, including the brain. And as the result the brain cells could not grow optimally.

In the other hand, adenoid hypertrophy reaching 90% of occlusion and macroglossia in this patient caused obstructive sleep apnea. Obstructive sleep apnea causes improper oxygenation during sleep yet during normal sleep, growth hormone is produced. And if patient did not get good sleep, in the morning and afternoon during the therapy, she will easily become fatigue. Thus, therapy would be inadequate.

Patient already suggested for adenoidectomy by the ENT department. And to prevent the obstructive sleep apnea, the patient was suggested to sleep in side lying positon or in prone position.

The limitations of this study is the short period of observation. The patient used the NGT after being told the usefulness nutrition for the child development.

In conclusion, the patient had Down syndrome with several comorbidities. The Down syndrome itself causes delay in development and the comorbidities make it worst. We need to find and manage the comorbidities, work together with other field so that our therapy could give the optimal result for the patient.

REFERENCES

Bruni, M. *et al.* 2010. 'Reported sensory processing of children with down syndrome', *Physical and Occupational Therapy in Pediatrics*, 30(4), pp. 280–293. doi: 10.3109/01942638.2010.486962.

van Dijk, M. and Lipke-Steenbeek, W. 2018. 'Measuring feeding difficulties in toddlers with Down syndrome', *Appetite*. Elsevier, 126(March), pp. 61–65. doi: 10.1016/j.appet.2018.03.018.

Ivan, D. L. and Cromwell, P. 2014. 'Clinical practice guidelines for management of children with down syndrome: Part II', *Journal of Pediatric Health Care*. Elsevier Ltd, 28(3), pp. 280–284. doi: 10.1016/j.pedhc.2013.05.003.

Kazemi, M., Salehi, M. and Kheirollahi, M. 2016. 'Down syndrome: Current status, challenges and future perspectives', *International Journal of Molecular and Cellular Medicine*, 5(3), pp. 125–133.

Kim, H. I. *et al.* 2017. 'Motor and cognitive developmental profiles in children with down syndrome', *Annals of Rehabilitation Medicine*, 41(1), pp. 97–103. doi: 10.5535/arm.2017.41.1.97.

Myrelid, Å. *et al.* 2002. 'Growth charts for Down's syndrome from birth to 18 years of age', *Archives of Disease in Childhood*, 87(2), pp. 97–103. doi: 10.1136/adc.87.2.97.

Wardah.2019. 'Sindrom Down'. Jakarta: Kementrian Kesehatan RI. Available at: http://www.depkes.go.id/resources/download/pusdatin/infodatin/infodatin down syndrom 2019.pdf.