IJCRT.ORG ISSN : 2320-2882

INTERNATIONAL JOURNAL OF CREATIVE RESEARCH THOUGHTS (IJCRT)

An International Open Access, Peer-reviewed, Refereed Journal

Growth and Development Analysis in Children with Trisomy 21 Down Syndrome

Aini Ariefa[1] & Irwanto[1]*

[1]Department of Child Health/Faculty of Medicine, Universitas Airlangga
Dr. Soetomo Academic General Hospital, Surabaya, Indonesia

Abstract

Down syndrome is one of the most common causes of intellectual disability and mental retardation in children. Moreover, Down syndrome children's motor development is slower than usual. Language development, especially expressive language, is significantly slowed, and the kid lacks a social orientation. This study aims to analyze growth and development in children with trisomy 21 down syndrome. Gender, age, mother age, paternal age, genetic factors, and physical traits of those with Down syndrome were all parameters used in this study. This study took place in the outpatient installation of Dr. Soetomo General Academic Hospital in Surabaya, assigning children aged 1 to 7 as sample. The SPSS application is used to process the data. Results show that out of 29 children with trisomy 21 down syndrome, 46.6% have less or very low weight, 66.6% have short or very short stature, and 90% have microcephaly. It was concluded that 21 trisomy down syndrome affect children's growth and development.

Keywords: down syndrome, children, growth disorder, developmental disorders.

Introduction

Several variables influence children's growth and development, including genetics (chromosomal anomalies), environment (prenatal, natal, postnatal), diet (macrosomal and micronutrients), stimulation, and hormones. Chromosomal disorders induce miscarriage, congenital deformities, and mental impairment [1]. With intellectual disability and physical development delays, down syndrome is one of the most common causes of intellectual disability and mental retardation in children. The condition was discovered in 1866 by a British doctor named John Langdon Down [2–4].

One of every 700 babies is born with down syndrome. Per some parts of the world, the incidence of down syndrome is 0.3–3.4 in 1000 births and accounts for about 25–30% of all mental impairment [5,6]. A doctor from Diponegoro University in Semarang, Indonesia, investigated a random sample of 123 patients with down syndrome for chromosomes, and their findings were discovered to be exact [7]. Having a kid with Down syndrome at the age of 30 carries a 1:1000 risk of having a child with the condition, but by the age of 40 the risk is 9:1000. The age of a woman when she is conceived significantly influences the likelihood of having a Down syndrome baby [4,7].

A variety of medical issues can be found in the majority of people with Down syndrome, including heart, endocrine, hearing, vision, bones and joints, hypotonia, and cancer. Moreover, Down syndrome children's motor development is slower than usual. Language development, especially expressive language, is significantly slowed, and the kid lacks a social orientation [4]. Down syndrome children have slanted eyes with a central angle forming a fold (epicanthal folds), small mouth (protusia), large tongue protruding (macroglossia), smaller head circumference (microchephaly), palms crossing straight/horizontally (simian

crease), and decreased muscle tone [8]. There have only been a few researchs on growth and development analysis in children with trisomy 21 Down syndrome in Surabaya up to this point.

Methods

This research is a cross sectional study, namely research conducted through observation or measurement at a certain time. This study is directed to analyze growth and development in children with trisomy 21 down syndrome. Measurement of risk factors in this study were maternal factors, paternal factors and genetic factors (heredity) on the incidence of Down syndrome. Both the risk factor and the effect, the measurement is only done once and at the same time. This research was conducted at the Pediatric Outpatient Installation of RSUD Dr. Soetomo from July to December 2019.

The population in this study was children with Down syndrome characteristics in the Outpatient Installation of RSUD Dr. Soetomo Surabaya. The sample of this research is children with Down syndrome characteristics aged 1 to 7 years in the Outpatient Installation of RSUD Dr. Soetomo Surabaya who met the inclusion criteria and did not get the exclusion criteria. The inclusion criteria of this study were: Children with physical characteristics of Down's syndrome, namely the presence of a mongoloid face, microcephaly, simian crease, epichantal folds, macroglossia, and a flat nose bridge, and were willing to participate in the study and signed an informed consent form. The exclusion criteria for this study were that parents were not willing to participate in the study and signed the informed consent.

The data collection instrument used in this study was a data collection sheet. Sources of data primary data are obtained from questionnaires and interviews. The data from each examination result is ensured to be complete and relevant prior to further processing. Re-examination is required before further management. The research data are presented in the form of tables, diagrams and text. The collection of research subjects was carried out at the pediatric outpatient installation of RSUD Dr. Soetomo Surabaya from July to December 2019. The subjects included in this study were children with Down syndrome who were taken by consecutive sampling. The diagnosis of Down's syndrome is based on the results of chromosomal analysis (karyotyping). Data entered in the data collection sheet (LPD) included gender, age, physical examination, chief complaint, birth history, family history and family diseases, growth history, social history and nutritional status. During the six-month study period, 33 children with Down's syndrome were found and were included as research subjects.

Univariate analysis in this study was conducted to describe each variable factor. Bivariate analysis using Chi-square test/Fisher's exact test, the test criteria are if the p-value < = 0.05 then the relationship is statistically significant, but if the p-value > = 0.05 then it is not statistically significant or not present. meaningful relationship. Data processing uses the Statistical Package for the Social Sciences (SPSS) program.

Results and Discussion

The number of samples that entered the inclusion criteria was 29 children, 16 male children and 13 female children. The risk factors for physical characteristics of the mongoloid face were associated with children with trisomy 21 Down syndrome (p=0.037). There is no correlation between gender, child's age, maternal age factor, father's age factor, family history, physical characteristics consist of macroglossia, simian crease and umbilical hernia to the results of chromosomal analysis. There were developmental disorders in children with trisomy 21 Down syndrome.

1. Basic Characteristics of Research Subject

The research subjects were presented based on basic characteristics, namely gender, age, maternal and paternal factors, heredity history, physical characteristics of Down syndrome children consisting of mongoloid face, macroglossia, simian crease and umbilical hernia. The basic characteristics of research subjects are presented in Table 1.

Table 1. Basic Characteristics of Research Subjects

Characteristics	Frequency	Percentage
Sex		
Male	16	55.2
Female	13	44.8
Age		
1-5th	14	48.2
>5th	15	51.8
Mother Factor		
Old age ≥ 35 years	18	62
Young age < 35 years	11	38
Father Factor		
Old age ≥ 35 year	21	72.4
Young age < 35 years	8	27.6
Genetic Factor		
There is a hereditary history	5	17.3
There is no a hereditary history	24	82.7
Mongoloid Face		
Yes	27	93.1
No	2	6.9
Macroglossia		
Yes	24	82.7
No	5	17.3
Simian Crease		
Yes	21	72.4
No	8	27.6
Hernia Umbilicalis		
Present	11	38
No	18	62

The number of patients with Down syndrome in the outpatient unit of RSUD Dr. Soetomo Surabaya during the study period of this research, namely from July-December 2019 were 33 patients. This number is 13% of the approximately 250 children with Down syndrome who are members of the East Java POTADS Foundation, and there are still many elementary school children who have not been recorded.

The results of the Ministry of Health's Basic Health Research data, in 2010, the prevalence of children with Down syndrome in Indonesia was reported to be 0.12% and increased to 0.13% in 2013 [9]. While the CDC records every year about 6000 babies are born with Down's syndrome or about 1:700 births of babies. From 1979 to 2003, the number of babies with Down's Syndrome increased by 30% [10].

In this study, 29 children with Down's Syndrome who met the inclusion criteria were included as samples. A total of 16 (55.2%) boys and 13 (44.8%) girls. Male predominance is common in Down's syndrome. This supports the hypothesis that the male sex ratio is higher in Down syndrome due to the failure of chromosome separation during spermatogenesis [11]. Research by Girirajan, 2009 revealed that due to meiosis disorders in the father that affect the sex chromosomes or at the time of fertilization, the accessibility of sperm carrying the Y chromosome is larger to the ovaries so that Down syndrome children born are male [12].

The mother and father factors aged 35 years in this study had a greater frequency, namely 18 (62%) and 21 (72.4%). This is in accordance with research by Stewart, which revealed that the incidence of Down syndrome is influenced by the age of the mother and father, where the higher the age of the mother during pregnancy, the greater the risk of giving birth to a child with Down syndrome [13]. Endocrine changes, such as increased androgen secretion, decreased hydroepiandrosterone levels, decreased systemic estradiol concentrations, changes in hormone receptor concentrations, and the hormones LH (Luteinizing Hormone) and FSH (Follicular Stimulating Hormone) which suddenly increase before and during menopause, can lead to a increase the likelihood of nondisjunction [2,13].

Father's age has an influence on Down syndrome children where parents of children with SD are found to be 20-30% of cases of extra chromosome 21 originating from the father. Aging of male spermatozoa cells and sperm maturation in the male reproductive organs are associated with a decrease in the frequency of intercourse and affect the extra chromosome 21 from the father [2].

In this study, 17.3% had a family history of children with Down syndrome. The chances of having a child with Down syndrome are higher if the pregnant woman has had a child with Down syndrome, or if there is a close family member who has a child with Down syndrome. There is an increased risk of recurrence if there is a child with Down syndrome in the family [13]

The characteristics of Down syndrome children in this study were mongoloid face 27 (93.1%), macroglossia 24 (82.7%), simian crease 21 (72.4%) and 11 children (38%) had umbilical hernia. This is in accordance with the physical characteristics of Down's syndrome children revealed by Devlin et al., namely children with Down's syndrome have physical characteristics, namely there is an angle in the middle forming a fold (epicanthal folds), a mouth that shrinks with a large tongue so that it looks protruding out (macroglossia), a relatively smaller head shape compared to normal people (microchephaly), palm tattoo that crosses straight/horizontally (simian crease), decreased muscle tone (hypotonia), flat nose bridge (depressed nasal bridge), short stature, hearing loss, chin smaller teeth (micrognatia) and teeth that are smaller than normal (microdontia) [14].

2. Growth Characteristics of Down Syndrome Children

Table 2. Down syndrome child growth characteristics

Development	Frequency	Percentage
Weight by age		
Weight is very low	5	17.2
Weight is less	9	31
Weight is normal	15	51.8
Height according to age		
Very short stature	6	20.7
Short stature	14	48.2
Normal stature	9	31.1
Weight to height		
Very bad	3	10.3
Short	9	31.1
Good	17	58.6
Head circumference for age		
Microcephali	27	93.1
Mesosephali	2	6.9

In this study, the growth characteristics based on weight according to age were found to have a normal weight of 15 children (51.8%). Children with Down syndrome had normal growth in weight-to-height measurements in 17 children (58.6%), short stature in 14 children (48.2%), and microcephaly in 27 children (93.1%). This is in line with research conducted in 2012 by Kawanto et al. at RSCM Jakarta on 60 children with Down syndrome. In this study, 60% of the children were good nutritional status, 90% short stature, and 75% microcephaly of the children studied [1].

Short stature was found in 14 children in this study, where short stature is a characteristic of most children with Down syndrome. The average height of most ages is around the second percentile of the general population [15]. Growth retardation in Down syndrome children has occurred since the prenatal period. After birth, the greatest decrease in growth rate occurs between the ages of 6 months and 3 years. Some of the conditions that cause growth retardation are congenital heart disease, thyroid hormone deficiency, celiac disease, upper airway obstruction, and nutritional deficiencies due to feeding difficulties. Puberty appears early and there is impaired growth support [16].

Research conducted by Arifiyah (2017) revealed that the assessment of height based on age was found to be mostly short stature of 46.5% and very short 24.4% of children, while the head circumference assessment was almost all microcephaly. The direction of the normal growth line in 75.6% of children [17]. According to Batubara, 2006 describes the growth curve of Indonesian children, the minimum average for children aged 6-12 years is 96-128 cm for girls and 96-125 cm for boys [18]. The average maximum height gain is 8.5 cm per year for boys and 7.3 cm per year for girls. The mean age at peak growth spurt was 12.3 years for boys and 10.8 years for girls, lower than healthy children [19].

The main cause of growth retardation is still unknown. Some conditions that can cause growth retardation are congenital heart disease, airway obstruction related to sleep apnea (sleep apnea), gastrointestinal disorders, thyroid hormone deficiency, and nutritional deficiencies due to feeding problems. There was also an increase in the incidence of overweight and obesity, especially in adolescence and adulthood. Regular monthly examinations in Down syndrome children can identify the causes of growth disorders and the risk of being overweight and obese. The growth curve is a useful tool for monitoring a child's growth. However, the growth pattern of Down's syndrome children differs from that of the general population, therefore it is important to use a specific curve for Down's syndrome [15].

3. The developmental characteristics of children with Down syndrome

The developmental characteristics of children with Down syndrome are presented in table 3. The developmental characteristics of Down syndrome children in this study were: Down syndrome children had motor development disorders 20 (69%), sensory development disorders 24 (82.8%), social development disorders 22 (75.9%) and language development disorders in 26 children (89.7%).

Table 3. Developmental characteristics of children with Down syndrome

Development	Frequency	Percentage
Motoric Development		
Normal	9	31
Not Normal	20	69
Sensory Development		
Normal	5	17.2
Not Normal	24	82.8
Social Development		
Normal	7	24.1
Not Normal	22	75.9
Language Development		
Normal	3	10.3
Not Normal	26	89.7

Conclusion

In summary, risk factor physical characteristics of the mongoloid face correlated with trisomy 21 Down syndrome children. There are developmental disorders that occur in children with trisomy 21 Down syndrome. The risk factors for the physical characteristics of the mongoloid face are related to children with Down syndrome clinically.

The weakness of this study is the limitation of the number of samples studied. However, this study can prove that there are growth and development disorders in children with Down syndrome. In the future, by knowing it early, it is hoped that delays in each aspect can be immediately identified and then followed up. Children with trisomy 21 need regular monitoring of growth and development in order to find out early if there is a delay in aspects of growth and development and can be identified for later follow-up.

References

[1] Kawanto F, Soedjatmiko. Pemantauan tumbuh kembang anak dengan Sindrom Down. Sari Pediatr 2007;9:185–90.

[2] Antonarakis S, Lyle R, Dermitzakis E, Reymond A, Deutsch S. Chromosome 21 and Down Syndrome: from genomics to pathophysiology. Nat Rev Genet 2005;5.

[3] Chandra N, Cyril C, Lakshminarayana P, Nallasivam P, Ramesh A, Gopinath PM, et al. Cytogenetic Evaluation of Down Syndrome: A Review of 1020 Referral Cases. Kamla Raj Enterp 2017;10:87–93. https://doi.org/10.1080/09723757.2010.11886090.

[4] Asim A, Kumar A, Muthuswamy S, Jain S, Agarwal S. Down syndrome: an insight of the disease. JBS 2015;22.

[5] Sommer C, Henrique-Silva F. Trisomy 21 and Down syndrome: a short review. Braz J Biol 2008;68:447–52. https://doi.org/10.1590/S1519-69842008000200031.

[6] Belmokhtar, Belmokhtar R, Kerfouf A. Cytogenetic study of down syndrome in Algeria: Report and review. J Med Sci 2016;36:46. https://doi.org/10.4103/1011-4564.181526.

[7] Nurhajjah S, Mariko R, Ramayumi R. Ekspresi Gen Superoxide Dismutase 1 serta hubungannya dengan fungsi timus penderita sindroma Down. Maj Kedokt Andalas 2014;34:1–5.

[8] Myrelid A, Gustafsson J, Ollars B, Annerén G. Growth charts for Down's syndrome from birth to 18 years of age. Arch Dis Child 2002;87:97–103. https://doi.org/10.1136/ADC.87.2.97.

[9] Riskesdas. Pokok-pokok Hasil Riset Kesehatan Dasar 2013. Badan Penelit Dan Pengemb Kesehat Kementrian Kesehat RI 2013:22.

[10] CDC. Data and Statistics on Down Syndrome. Natl Cent Birth Defects Dev Disabil Centers Dis Control Prev 2019.

[11] Kovaleva N, IButomo I, Körblein A. [Sex ratio in Down syndrome. Studies in patients with confirmed trisomy 21]. Tsitologiia i Genet 2001;36:43–9.

[12] Girirajan S. Parental-age effects in Down syndrome. J Genet 2009;88:9–14.

[13] Stewart K. Trisomy 21-Down's syndrome. Australas. Genet. Resour. B., 2007.

[14] Devlin L, Morrison P. Accuracy of the clinical diagnosis of Down syndrome. Ulster Med J 2004;73:4–12.

[15] Styles M, Cole T, Dennis J, Preece M. New cross sectional stature, weight, and head circumference references for Down's syndrome in the UK and Republic of Ireland. Arch Dis Child 2002;87:104–8. https://doi.org/10.1136/ADC.87.2.104.

[16] Wasant P, Boonyawat B, Tritilanunt S, Vatanavicharn N, Sathienkijakanchai A, Ratanarak P. Factors in uencing development of Down syndrome children in the first three years of life: Siriraj experience. J Med Assoc Thai 2008;91:1030–7.

[17] Arifiyah, Asri P. Hubungan antara Insulin-like Growth Factor-1 dengan Pertumbuhan dan Perkembangan Anak Sindrom Down. Sari Pediatr 2017;18:350–6.

[18] Batubara J, Alisjahbana A, Gerver-Jansen J, Alisjahbana B, Sadjimin T, Tasli Y. Growth diagrams of Indonesian children the nationwide survey of 2005. Paediatr Indones 2006;46:5–6.

[19] Diamond K, Huang H, Steed E. Chapter thirty-three: the development of social competence in children with disabilities. 2007.