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Original Article

Developmental assessment of children with Down syndrome

Serpil Dincer[1], Derya Gumus Dogan[2], Meltem Kivilcim[3], Sinem Kortay Canaloglu[2]

[1]Dr. Lutfi Kirdar Training and Research Hospital, Department of Paediatrics, Istanbul, Turkey
[2]Inonu University Faculty of Medicine, Department of Pediatrics, Division of Developmental and Behavioral Paediatrics, Malatya, Turkey
[3]Behcet Uz Training And Research Hospital, Department of Pediatrics, Division of Developmental and Behavioral Paediatrics, Izmir, Turkey

Abstract

Aim: Morbidity and mortality of children with Down syndrome (DS) have decreased and their quality of life has increased with modern medicine. The purpose of this study is to assess the developmental characteristics and activities of children with DS and also their participation to life, the environmental factors, the services they receive and their health and sociodemographic characteristics by using Expanded Guide for Monitoring Child Development (E-GMCD)

Material and Methods: Children diagnosed with DS who were admitted to hospital were assessed with E-GMCD. Health information and the family's sociodemographic features were supplied from families and hospital files.

Results: The study included a total of 100 children diagnosed with DS, 41 girls and 59 boys, who were between 2-59 months of age. The mothers of the 63 children stated that they were concerned about their children's learning, motor skills, communication, relationships with others, and senses (sense of hearing and seeing). Of the 57 (81%) children were found to have delay in expressive language and were older than 1 year old ($p<0.05$). The delay in gross motor domain seemed to decrease with the increase by age, it was not found to be statistically significant ($p>0.05$). A majority of children who received specialized education and physiotherapy were between ages of 2 and 4.

Conclusion: Children with DS should be monitored regularly beginning from the day they are born. These children should get early education to speed up their cognitive development. Family centered early support programs should start as early as possible.

Keywords: Down Syndrome; Developmental; Assessment.

INTRODUCTION

Down syndrome (DS, OMIM #190685) has a prevalence of 1 in 691 live births and it is the most prevalent genetic cause of intellectual disability (1). It is known to be correlated with a great number of other medical conditions. Children with DS have higher prevalence of cardiovascular, gastrointestinal and sleeping disorders. Autism, epilepsy, intellectual disability, ear diseases, eye disorders, hypothyroidism, diabetes, and obesity are commonly high in DS patients regardless of age (2).

Due to their susceptibility to certain medical problems, children with DS require careful medical care. As soon as the diagnosis of DS is made, an assessment process should be started to settle the appropriate interventions for all developmental domains (3). People with DS should be actively monitored in terms of health all throughout their lives; in addition, preventive health screening and diagnostic practices should be developed and implemented (4). In clinical practice and research, the most appropriate tool that could be used should include a child's functional experience, ability to participate and the environmental context in which the child lives (5). In terms of health and environment, the most extensive model to describe human functioning was adopted by WHO in 2001 (6) and it was adapted for use in children and adolescents in 2007 (Child and Youth version: ICF-CY) (7). International Classification of Functioning does not only limit an individual's health with the basics of the etiology of a disease; but also makes it possible to classify the processes about a person's functions, activities and participation in many areas of life and also to classify the environmental factors that can affect all these characteristics.

The purpose of this study is to determine the functions,

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Corresponding Author: Derya Gumus Dogan, Inonu University Faculty of Medicine, Department of Pediatrics, Division of Developmental and Behavioral Paediatrics, Malatya, Turkey, **E-mail:** deryagumus@yahoo.com

activities and life participation of children with DS and to determine the environmental factors which affect these features of the child based on the ICF-CY model by using Expanded Guide for Monitoring Child Development (E-GMCD).

MATERIAL and METHODS

One hundred children with DS under the age of 60 months who were admitted to Inonu University, Faculty of Medicine, Developmental and Behavioral Pediatrics Polyclinic and who had diagnosis through chromosome analysis were included in the study. Following the implementation of GMCD to patients who met the inclusion criteria, the child's health information and the family's sociodemographic features were supplied from families and hospital files.

GMCD and E-GMCD

Expanded Guide for Monitoring Child Development (E-GMCD) is the expanded form of Guide for Monitoring Child Development (GMCD) developed by Ertem et al. which also covers ICF-CY (8).

The first part of E-GMCD includes sociodemographic information.At the end of the first part, the family is asked whether they are concerned about the development of their child. The second part includes GMCD. This part assesses the child's expressive and receptive language, gross and fine motor skills, and relationships with the environment, play and self-care characteristics. GMCD is a short, easily administered tool which provides the participation of the family in the assessment of development of infancy and early childhood period (9). The third part includes questions about the child's developmental functions, activities and life participation. The fourth part questions the environmental factors that may affect the child. The fifth part includes services about the child's development. The sixth part includes services about the child's health. The seventh part includes prenatal, birth and newborn information and family history while the eighth part is about the physical examination, laboratory and other examinations. The ethical board approval of the research was taken from Ethical Board of Inonu University.

Statistical Analysis of the Data

The statistical analysis of the data was made by using "StatisticalPackageforSocial Sciences (SPSS 17)"packageprogram. The children who formed the sample were analyzed in terms of descriptive statistics (frequency, average/mean and distributions). The differences between groups were assessed by Chi Square statistical test. A difference was considered between groups when the p value was under 0.05.

RESULTS

Sociodemographic Features

Of the 100 children, 59 (59%)were boys while 41 (41%) were girls. Average age of the children was 24±14.9 months. One child had Robertsonian translocation (14;21), 2 children had mosaic karyotypes and the rest had trisomy 21. The percentage of births that were preterm was 24%. Over 28% of all children had low birth weight. Table 1 shows the sociodemographic characteristics and newborn information of the children who formed the sample.

Table 1. Sociodemographic characteristics of children with Down Syndrome

Sociodemographic Characteristics	n=100 (%)
Gender	
Girl	41 (41)
Boy	59 (59)
Child age (months)	
<13	31 (31)
12-25	24 (24)
26-35	19 (19)
36-49	21 (21)
50-61	5 (5)
Maternal age (years)	
18-25	16 (16)
26-35	37 (37)
36-49	47 (47)
Maternal education	
Illiterate	14 (14)
< 8 years	63 (63)
> 8 years	23 (23)
Gestational Age, weeks	
>37	76 (76)
≤ 37	24 (24)
Birth weight, g	
2500gram -4000	68 (68)
<2500	28 (28)
> 4000	4 (4)

Of the 100 children, 45 had only cardiac problems, 23 had endocrine problems, 3 had neurological problems and 1 had AML diagnosis. One patient had gastrostomy, 12 children had both hormonal and cardiac problems. Of the 57 children who had congenital heart disease, 25 (41%) had atrial septal defect, 13 (22%) had ventricular septal defect, 15 (25%) had atrioventricular septal defect and 4 had (6%) patent ductus arteriosus and 25 (41%) had undergone cardiac operation.

Frequency of developmental problems

Of the families 63 (63%) had stated that they were concerned about the development of their children. Of the patients who had concerns, 75% were concerned about motor skills, 73% about language, 25% about relationships, 10% about games and 9% about self-care skills while 65% were concerned about the development of both motor skills and language. Twenty-one (21%) children were found to have delays only in one area while 67 (67%) were found to have delays in more than one area. The number of children who did not have delays in

any area was 12(12%). Sixty percent of the children who were not found to have any delays in the developmental stages were children with DS who were under the age of 1 and this result was statistically significant (p<0.05). The results of the assessment of the children's development through GMCD are given in Table 2.

Expressive Language: Seventy (70%) children were found to have delays in expressive language, 13 (8%) of the children had delays in receptive language before the age of 1, while 54 (82%) were found to have problems in receptive language after the age of 1. Delays in expressive language are in the lowest level in the first twelve months.

Receptive Language: While 36 (36%) children were found not to have any problems, 64 (64%) children were found to have experience delays in this area. Ten (16%) children before the age of one had delays in receptive language while 54 (54%) children after the age of one had delays in receptive language and this result was statistically significant (p<0.05).

Gross and Fine Motor: Fifty two (52%) children were found to have delays in their gross and fine motor domains. Of the children who had delays, 15 (25%) were within their first year of age, 19 (31%) were between the ages 1-2, 13 (21%) were between the ages 2-3, 12 (20%) were between the ages 3-4 and 2 (3%) were between the ages 4-5.

Relationship:Seventy-six (76%) of the children were found to have positive relationships with the environment such as making eye, contact knowing their mothers and relatives and showing their feelings, being able to make others participate in their games and trying to attract attention.

Play: In the domain of play, 27 (27%) children were found to have delays.

Self-care:Of the69 children older than one, 39 (56%) were found to have developmental delays (such as eating and getting dressed) in self-care skills.

Developmental stages of the children with congenital heart disease

Of the 57 children with DS who had congenital heart disease, 41 (71%) were found to have delays in expressive language, 40 (70%) in receptive language, 37 (64%) in gross motor skills, 13 (23%) in relationship and 18 (32%) in play domain. Forty children older than 12 months of age were assessed in terms of self-care skills and 24 (60%) were found to have delays. Ten (18%) of these children had delays only in 1 area, 36 (63%) had delays in more than one area and 11 (19%) were not found to have delays in any area.

Assessment of developmental functions, activities and life participation

Of the families 37 (37%) stated that they did not experience any problems in terms of learning skills, attention and interest. When the options of shy, noncompliant, sad, timid, uncomfortable, still and calm were assessed as negative and the options of happy, brave, cooperative, curious and friendly were assessed as positive, 31 (31%) families stated that they had positive characteristics. Table 3 presents information about learning, attention, sleep and feeding status of the children declared by families.

Table 2. The developmental status of children according to E-GMCD

	Expressive Language Skills n=100 (%)	Receptive anguage Skills n=100 (%)	Gross and Fine Motor Skills n =100 (%)	Relationship n=100 (%)	Play n=100 (%)
Children with developmental delay	70 (70)	64 (64)	61 (61)	76 (76)	27 27)
Children without developmental	30 (30)	36 (36)	39 (39)	24 (24)	73 (73)

Table 3. Attention, Learning, Feeding and Sleep Status of Children

	Attention, Learning n=100 (%)	Sleep Status n=100(%)	Feeding Status n=100(%)
Children with problem	18 (18)	8 (8)	15 (15)
Children with a little problem	45 (45)	23 (23)	21 (21)
Children without problem	37 (37)	69 (69)	64 (64)

Environmental

Parents' spending time with their children, playing games and talking with them were assessed as positive things done by families to support the learning and development of their children and no problem was found in 89 (89%) of the children.

When the parents were asked about their supports on "moral and material support for the development of their children", 35 (35%) stated that there were problems.

Services about the children's development and health

It was found that 51 (51%) of the children had not received specialized education yet. Average age for starting specialized education was found as 19±10 months. Of the children who received specialized education, 2 (4%) were within their first year, 11 (22%) were between 1-2 years of age, 14 (29%) were between 2-3 years of age, 17 (35%) were between 3-4 years of age and 5 (10%) were between 4-5 years of age. Of the children who received specialized education, 14 (29%) were girls, while 35 (71%) were boys, 92% had started before 42 months. Thirty-five (35%) of the families stated that their children received only physiotherapy while 28 (28%) stated that their children received both physiotherapy and specialized education. Average age for receiving physiotherapy was

found as 27±60 months. Of the parents of 49 children who received specialized education, 39 (80%) stated that they were pleased about the education their children received, 5 (10%) families stated that they were not pleased and 5 (10%) families stated that the education was not sufficient.

While 73 (73%) children received regular health services, 22 (22%) children did not receive regular health services. It was found that 5 (5%) children did not receive any health service at all. Average hospitalization number of children with Down syndrome was 3. Of 100 children, 37 (37%) had been hospitalized more than five times with various reasons. Twenty-six (72.9%) of these consisted of children with congenital heart disease. Of the children 84% who had been hospitalized more than five were found to have delays in the stages of development.

DISCUSSION

This study defines the developmental, functional, social and environmental profiles of a group of young children with DS according to ICF-CY.More than half of the families are concerned about the development of their children. Most children have developmental delays, especially after one year old. Half of the children had not received any special education. The average age for starting specialized education is very late. Children with DS, mostly those having CHD are hospitalized frequently.

The delay in expressive language is more obvious than the delay in receptive language in children with DS. Progress in expressive language tends to take place later when compared with receptive language and cognitive skills. In a study conducted with 1620 families who had children with DS, Kumin et al. found that children with DS understood more than they could tell. It is thought that this may be the result of hearing loss, midface hypoplasia, medium size tongue and lips and low muscle strength of the tongue (10,11). In children with DS, an early language-based communication goal making use of total communication (objects, pictures, signs, gestures, printed and spoken words) is essential due to relative strength in the receptive language skills compared with expressive language skills (12). Gross motor skills such as walking and running are acquired later when compared with children with typically developing children (13- 15). In our study, half of the children who were found to have delays in their motor domains were within the first two years of their lives. In a study which compared the gross motor skills of children with Down syndrome with typically developing children in their second, fourth and twenty-fourth months, it was found that children with DS had difficulties in activities which required speed, postural control, balance and early postural control (16). In children with DS, motor performances are more delayed between 7 and 12 months than between 3 and 7 months (17), and more delayed at 36 than at 18 months of age (13). Children with DS are shown to be responsive to specific training in these types of gross motor skills (15,18).

More than half of the children had CHD. Karaman from Turkey had declared that the prevalence of CHD in children with DS was 22.4% in his study group (19). The majority of children with developmental delays are those with CHD in our study. In a study patients with DS who had AVSD and those who did not have AVSD were compared, greater developmental deficits were found in the motor domain in children with CHD (12).

In children with DS, problems in sleep start at an early age and may continue with increasing age. Toddlers who had DS and poor sleep (66%, n = 19) were shown to have greater deficits on parent-reported and also objective measures of language, including vocabulary and syntax (20). In Bassell's study 76% of the caregivers who had children with DS stated that their children had sleep problems between the ages of 1.50 to 13.4 years (21). Due to their hypotonia, relative macroglossia, small oral cavity, increased risk of oral-motor dysfunction (dysphagia), increased incidence of sensory issues, and developmental problems, infants and children with DS are at high risk for feeding challenges (22).

In this population, about 80% of hospitalizations and intensive care unit admissions occur due to respiratory infections (23). As the number of hospital admissions increase, the rates of developmental delay also increase. Congenital heart and gastrointestinal disease and acquired respiratory disease cause high rates of hospital admission and medication use rates in young infants with DS (24). In our sample, delay in developmental stages was found in 84% of the children who had more than five hospitalization history.

There is limited research about DS children through ICF-CY. There are two studies mentioning ICF-CY similiar to our study. But in those researches the age of the children are older than our group. Jung et al compared function, activity and quality of life between DS children and typically children who were between 4-12 years old. Statistically significant differences were found between two groups. DS children showed low activity and participation (25). MacDonald et al. investigated the participation patterns of 62 children between the ages of 9 and 17 years with (DS). It was found that children with DS participate more in informal compared to formal activities (26).

Children with DS can benefit greatly from specialized education and interventions (27,28). They can benefit more from early education and rehabilitation programs when compared with other children with mental retardations (29). In our study, only half of the children with DS received specialized education and the average age for starting specialized education is 19 months, which is too late. With the diagnosis of DS, it is crucial to initiate assessments for the determination of appropriate interventions to address all developmental domains. This process should begin at the time of diagnosis. It is unnecessary to wait for a developmental delay to initiate an ongoing assessment and intervention process (3).

CONCLUSION

During the last fifty years, life expectancy of individuals with DS has increased greatly due to advances in medical care. The health and well-being of these individuals are

improved by sufficient healthcare (30). These children with intellectual disability and various organ dysfunctions should be assessed according to biopsychosocial model. American Academy of Pediatrics (AAP) has published guidelines for the health care of children with DS to help pediatricians in preventative medical care (31). Health personnel working with children who have DS should monitor these children within the context of domains defined by ICF-CY and in line with the suggestions of AAP.

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Serpil Dincer ORCID: 0000-0003-0653-6546
Derya Gumus Dogan {ORCID:0000-0003-1587-9639
Meltem Kivilcim ORCID: 0000-0001-6305-6835
Sinem Kortay Canaloglu ORCID: 0000-0003-4733-0181

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