Original Article

Developmental Profile as a Predictor of Behavior Phenotype in Down Syndrome Children

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ABSTRACT

Objective: To study the Emerging Down Syndrome Behavioral Phenotype from infancy to Early Childhood and to see Developmental Profile as a Predictor of this Behavior phenotype in Down Syndrome Children.

Study Design: Cross-sectional study

Place and Duration of Study: This study was conducted at the Department of Development and Behavioral pediatrics, University of Child Health science, Children's hospital Lahore and duration of study was 6 months from 1st January 2023 to 30th June 2023.

Methods: It was a Cross-sectional study. Data was collected from Department of Development and Behavioral pediatrics, University of Child Health science, Children's hospital Lahore and duration of study was 6 months. A sample of 42 Children having age 1-5 years was collected through purposive sampling technique.

Results: In breakup of PEEP and SDQ tools, Cognition age of 9 (19%) patients was profound, 23 (48%) severe, 9 (19%) moderate and 7(15%) was mild delay. Socialization age of 4 (8%) patients was profound, 11 (23%) severe, 13 (27%) moderate, 11(23%) mild and 9 (19%) patients were age appropriate. Self-help age of 7(15%) patients was profound, 12 (25%) severe, 13 (27%) moderate, 10 (21%) was mild delayed and 6 (13%) patients were patients were age appropriate. Motor age of 8(17%) patients was profound, 13 (27%) severe, 15 (31%) moderate, 8 (17%) was mild delay and 4 (8%) patients were age appropriate. Expressive Llanguage age of 19 (40%) patients was profound, 22 (46%) severe,5 (10%) moderate 1(2%) was mild delay and 1 (2%) patients were age appropriate.

Conclusion: Specific Behavioral Phenotype (BF) in children with Down Syndrome (DS) is consistent with relative strength in sociability, non-verbal abilities, receptive language, implicit memory skills and visuo spatial processing and relative weakness in gross & fine motor skills, verbal communication, visuo motor, cognitive functioning and motor planning.

Key Words: Down syndrome (DS) Behavior Phenotype (BP), Development Profile (DP), Portage Early Education Plan (PEEP)

Citation of article: Abid MB, Zahra SW, Kamran M, Munir SB, Ehsan Ullah, Ghous M. Developmental Profile as a Predictor of Behavior Phenotype in Down Syndrome Children. Med Forum 2024;35(1):87-91. doi:10.60110/medforum.350120.

INTRODUCTION

Down syndrome (DS), trisomy 21, is the most common genetic disorder, with an estimated incidence of 1 in 700 live births[1]. Out of all genetic disorders, a lot of research has been done on Down syndrome. Development is typical in infancy, slows down in next two years amid delayed rate of brain myelination.

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Received: December, 2023
Accepted: December, 2023
Printed: January, 2024

The Down syndrome (DS) behavior phenotype has been described as having relative strengths in non-verbal receptive skills and sociability & relative weakness in cognition expressive language and motor planning[2]. The most constant and typical features of DS are intellectual disability and craniofacial dysmorphism[3], with a variety of organ involvement and systemic diseases[4]. By definition behavioral phenotype refers to observable characteristics that occur more often in individuals with a specific genetic syndrome than individuals without that syndrome[5]. Characters that are not observable are called endo-phenotype and include thoughts, emotions and motivational states. Behavior phenotype in children with DS has typical characteristics and is explained in domains of his/her cognition/intellect, socialization, speech and language communication, self-help and motor skills which are collectively called as developmental profile[5] .

In cognition domain of development, children with DS have mild to severe delay with the profile of relative strength and weakness. They tend to have difficulties

more in understanding & following commands impaired memory, learning & self-care but they are good in non-verbal abilities and visually stored memory[6].

As regards to Speech and Language domain of development children with DS have better receptive than expressive skills in the verbal domain[7]. Receptive vocabulary can be considered as a relative strength. The understood more than they can speak. As for the expressive domain, children with DS have been found weak in phonology, grammar and syntax, while their intentional use of communication and gestures, and their social use of communication generally seem to be in line with their mental age[8]. A pattern of strengths and weaknesses has been observed in the nonverbal domain as well. In terms of social development children with DS are relatively strong and often described as charming affectionate, outgoing, cheerful, happy and sociable. Motor functioning demonstrates specific motor impairments in a number of fine and gross motor tasks (i.e., balance, posture, strength, and flexibility), as well as motor planning (i.e., praxis), although CA-level performance has been observed in specific skills including, running speed, agility, and visual-motor control. Conductive Hearing impairment negatively impacts language development[9].

This research will help in establishing the developmental profile in early childhood can be used as a predictor of Behavior Phenotype (Strengths and weakness) in children with Down syndrome.

METHODS

It was a Cross-sectional study. Data was collected from Department of Development and Behavioral pediatrics, University of Child Health science, Children's hospital Lahore and duration of study was 6 months. A sample of 42 Children having age 1-5 years was collected through purposive sampling technique. PEEP (Portage Early Educational Plan) Guide was administered to determine developmental Delay in 5 domains of development (Cognition, Self Help, Socialization, Motor and Speech). Strength and Difficulties Questionnaire (SDQ) was incorporated to exclude children with DS having behaviors related to pervasive development disorders. Demographic Questionnaire including Age, Gender, Family Size, Education, income etc. was also administered.

RESULTS

Data was analyzed using SPSS 25.0. Demographic and socio-economic data of our research shows that, 36 (75%) patients were male and 12 (25%) patients were female. 13 (27.1%) patients were belonging to nuclear family system and 35 (72.9%) patients were belonging to joint family system. 30 (62.5%) parents of patients have low socio-economic status, 16 (33.3%) parents of patients have middle socio-economic status and 2 (4.2%) parents of patients have high socio-economic status. 40% father and 44% mothers of patients were un-educated.

Figure 1 shows that, in breakup of PEEP and SDQ tools, Cognition age of 9 (19%) patients was profound, 23 (48%) severe, 9 (19%) moderate and 7(15%) was mild delay. Socialization age of 4 (8%) patients was profound, 11 (23%) severe, 13 (27%) moderate, 11(23%) mild and 9 (19%) patients were age appropriate. Self-help age of 7(15%) patients was profound, 12 (25%) severe, 13 (27%) moderate, 10 (21%) was mild delayed and 6 (13%) patients were age appropriate. Motor age of 8(17%) patients was profound, 13 (27%) severe, 15 (31%) moderate, 8 (17%) was mild delay and 4 (8%) patients were age appropriate. Expressive Language age of 19 (40%) patients was profound, 22 (46%) severe, 5 (10%) moderate 1(2%) was mild delay and 1 (2%) patients were age appropriate.

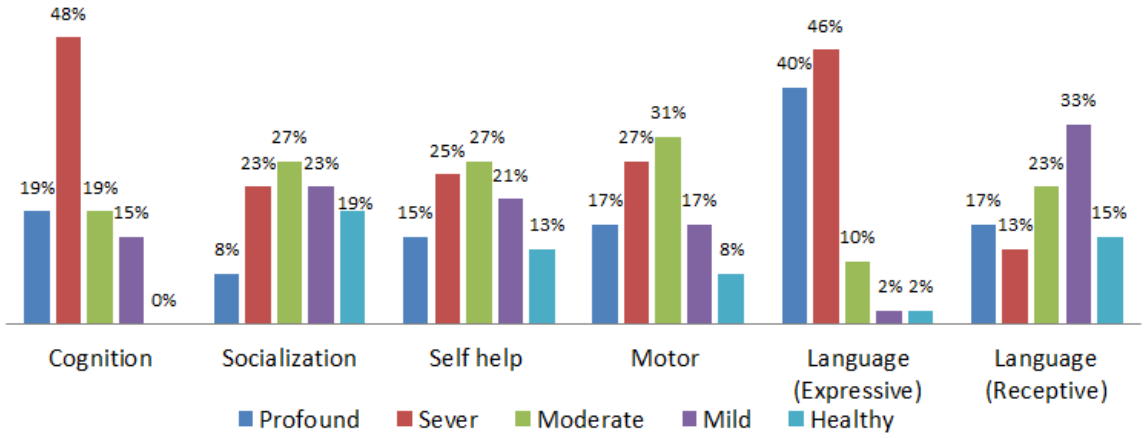
Table 1: this table shows, throwing 34 (71%), Teasing 5 (10%), Screming 32 (67%), Selfdirected 16 (33%), and Directed toward others 34 (71%). Table 2: shows that average age of patients was 35.79 months, Cognition age was 16.61 months, Socialzation age was 27.43 months, Fine Motor and Self-Help age was 21.96 months, Gross Motor age was 21.13 months, Receptive language age was 28.18 months and Expressive language age of Down syndrom (DS) patients was 12.73 months. Table 3: shows that there was a significant difference between choronoligcal age and socialization age, choronoligcal age and self-help age, choronoligcal age and Gross Motor age, choronoligcal age and Receptive Language age, choronoligcal age and Expressive Language age of DS patients.

Table No.1 : Secondery Behaviuoral problems

Behaviors of individuals	Yes	No
Throwing	34 (71%)	14 (29%)
Teasing	5 (10%)	43 (90%)
Screaming	32 (67%)	16 (33%)
Self-directed	16 (33%)	32 (67%)
Directed toward others	34 (71%)	14 (29%)

Table No.2: Paired sample statistics

Age	Mean	Std. Deviation
Chronological age of patients	35.79	15.12
Cognition age	16.61	11.19
Socialization age	27.43	11.97
Fine Motor and Self-Help age	21.96	13.31
Gross Motor age	21.13	13.15
Receptive Language age	28.18	9.13
Expressive Language age	12.73	10.20



Figurer No.1: Cognition age, socialization age, self-help age, Motor age and language age of patients

Table No.3: Output of Paired sample t-test

Variables	Paired Differences		T	p-value
	Mean	SD		
Chronological age - Cognition age	19.18	10.958	12.124	<0.005
Chronological age -Socialization age	8.37	10.623	5.460	<0.005
Chronological age - Fine Motor and Self-Help age	13.83	11.360	8.435	<0.005
Chronological age – Gross Motor age	14.66	9.664	10.509	<0.005
Chronological age – Receptive Language age	23.07	9.160	17.447	<0.005
Chronological age - Expressive Language age	23.07	11.810	13.532	<0.005

DISCUSSION

Specific Behavioral Phenotype (BF) consistent in children with Down Syndrome (DS) presented relative strength in sociability, non-verbal abilities, receptive language, implicit memory skills and visuo spatial processing and relative weakness in gross & fine motor skills, visuo motor, cognitive functioning, expressive language, communication and motor planning. This BF is clearly related to the development profile (DP) of the child and early development of typical behavioral characters (BC). We measured the DP & BC through PEEP and SDQ tool was incorporated to exclude children with pervasive development disorders and found PEEP as predictor of typical emerging BF in DS children.

Previous studies have reported this specific behavioral phenotype in children above 5 years[10]. This study was shaped to see this BF through developmental & behavioral assessments of children with DS before 6 years. Description and results of the research is viewing behavioral phenotypes from a developmental perspective. It seemed that this particular phenotype appears in early years of development and then emerge slowly before five years of age.

The Expressive Language (EL) was assessed through child's spontaneous answers, spoken responses to questions and multi word sentences. The expressive language words were all centrally processed, requiring the child to respond to an auditory input. Expression was in line with comprehension and auditory processing and mechanical motor skills. Expressive Language (EL) skills were recorded on parents information and direct conversation with the child and noted according to number of intelligible words, 2 or 3 words speech and sentence formation these findings are agreement with previous studies [11, 12].

A specific cognitive profile was observed in children with (DS) showing difficulties in intelligible and comprehensive language and spoken memory challenges, and relatively stronger non-verbal abilities and visual memory skills. They are better in receptive than expressive skills in the verbal domains, meaning thereby that these children understood more words than they are able to speak. Receptive Vocabulary (RV) is relatively better, but the depth and breadth of their RV is weak. DS children have been found weak on phonology, grammar and syntax but their social cues and gestures are generally according to their mental age. Low cognition has been ascribed due to deficits in verbal processing, large differences in expressive and receptive language domains and smaller size of intelligible spoken words. Our study showed cognitive age as 16.61 months while other study[13] showed it as 18.22 months.

The communication domain involved receptive and expressive language. In communication competence, non- verbal joint attention and gestural language are better whereas non-verbal requesting behavior showed deficits in children with DS. In receptive language

child's verbal input is determined as the small item questions they can understand better like pointing and simple commands but complex items involving multi tasks in one command were difficult to perceive. In our study Receptive and Expressive language of patients were 28.18 and 12.73 months respectively while previous study[13] showed that Receptive and Expressive language as 22.11 and 19.33 months respectively.

The Gross Motor (GM) functions were assessed as at what age child developed head control, turning sides, and prone to spine, sitting, crawling, standing in a manner to know progress in cephalocaudal and proximodistal motor development. In preschool children GM skills like running, hopping, jumping, climbing up & down stairs and tricycle riding. ("Pedals tricycle or other three-wheeled vehicle for at least six feet") were noted as their age of development. Our study showed Gross Motor age as 21.13 months while older study[13] showed it as 18.22 months.

The Fine Motor (FM) and Self Help skills involved bilateral and unilateral hand functioning. Bilateral items included closing zip, opening book and turning pages, buttoning/unbuttoning, lacing shoes, sharpening pencils, wearing socks & shirts, use of cloth clips, folding, and cutting etc. Unilateral items included eating with sticks, use of spoon and forks, holding feeder, bridging with blocks, marking tower of blocks, use of peg board, etc. Our study showed Fine Motor age as 21.96 months while older study[13] showed it as 20.83 months

The Socialization Domain assessed through functioning in peer relationships (PR), play and entertainment hours, and adapting skills. For toddlers, PR items involve like "laughs or smiles appropriately in response to positive statements. For preschool children dimensions like "participates in at least one game or activity with others", and coping skills involve items like "says 'please' when asking for something". Age equivalent scores for the Down syndrome group in this study on Play and Leisure Time socialization averaged 27.43 months, in contrast to the previous study[13] the (PR) area domain showed average age equivalent scores of 20.17 months. Positive emotions signals & smile frequencies were also found high.

In this context two marked differences were noted in individuals with Down syndrome. It was small at early developmental ages as regards to difference between expressive and receptive language, which averaged only 2.5 months. Whereas, it was large with an average of 20 months in older children with Down syndrome. That small dissociations early in development can result in increasingly larger differences over time is consistent with dynamic systems theory in those small starting state differences can evolve considerably larger as development becomes increasingly complex and differentiated[14,15.]

The Visual Reception measures were taken from visually stored memory. Children were exposed to various objects in different forms and shapes which involved oculomotor nerves and central visual pathways in localizing single and multiples points on surface through visual tracking[16].

CONCLUSION

In the light of new understanding of development in genetic syndromes and as a part of larger movement towards studying BF particularly in children with DS, it has become possible now to focus on weaker areas of BF like expressive language and motor skills before they become areas of pronounced weakness. Areas of relative strength may be taken as" gateway in" to polish areas of weakness to prevent future delays by early identification of areas of strength & weakness through developmental profile in toddler years taking as potential windows of opportunity to address weaker areas of BF is in toddler age group before they become pronounce in preschool years.

Visuospatial aspects of visual recognition memory, visual motor integration and visual imitation as areas of strength in children with DS are useful sub domains to enhance cognition level in these children.

Author's Contribution:

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Revisiting Critically:	M Bilal Abid, Syeda Wajeeha Zahra
Final Approval of version:	M Bilal Abid

Conflict of Interest: The study has no conflict of interest to declare by any author.

Source of Funding: None

Ethical Approval: No.752/CH-UCHS dated 27.12.2022

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