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Original Article

Sociodemographic and Clinical Profile of Down Syndrome Children Less Than 18 years and Its Effect on Caregiver Burden in a Tertiary Care Center

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Abstract

Introduction: *Down syndrome is one of the leading causes of intellectual disability. They face various health issues, including speech abnormalities, congenital heart diseases, Alzheimer's disease, leukaemia, and Hirschsprung. The emotional and social stress that these parents undergo because of such a child in a family is immense. This study aims to determine the sociodemographic and clinical profiles of children with Down syndrome.*

Objective: *To assess the sociodemographic and clinical profile and caregiver burden of children with Down syndrome.*

Methodology: *Data is obtained by personal interview using a standard proforma and consent from caregivers attending the Regional Early Intervention Centre and the Department of Paediatrics at Government TD Medical College, Alappuzha. Subjects were interviewed using a questionnaire that includes a burden interview, sociodemographic profile, Kuppswamy scale, modified Zarit scale for caregiver burden, and a clinical examination.*

Results: *57.5% are men. Mother primarily belongs to the 18-25 age range. Paternal education level is higher than maternal. The majority of fathers work in semi-skilled jobs, while most mothers are homemakers. Upper lower-class families have more. The majority resides in rural areas. In most cases, the mother serves as the primary caregiver. Most of them attend special school. The majority bear a mild to moderate burden. Anthropometry is normal in the majority. Hypothyroidism affects 50% of people. Mild intellectual disability is more. OSASD is a common CHD.*

Conclusion: *Males are more. Maternal age is younger during pregnancy. Paternal education and occupation rank higher than maternal. Upper-class families are more common, and the majority of the study population lives in rural areas. In most cases, the mother serves as the primary caregiver. The majority of the pupils attend special school. Mild to moderate burden is prevalent. Hypothyroidism affects 50% of people. Mild ID is more. OSASD is a common CHD.*

Keywords: *Down syndrome, clinical profile, sociodemographic profile, caregiver burden*

Introduction

Down syndrome is the most frequent chromosomal disorder, with an incidence of 1/600 to 1/1000 live births worldwide. It results from chromosome 21 trisomy, and there is a higher risk associated with advanced maternal age. The presence of an intellectual disability occurs with varying degrees in these individuals.(1) A mentally retarded child in a family is usually a serious stress factor for parents and often requires reorientation and re-evaluation of family goals, responsibilities, and relationships.(2)

Down syndrome is associated with congenital anomalies and characteristic dysmorphic features. The constellation of phenotypic features is fairly consistent and permits clinical recognition of trisomy 21. Phenotypic features include cleft lip, flexed fingers with postaxial polydactyly, ocular hypotelorism, bulbous nose, low-set malformed ears, microcephaly, cerebral malformation, especially holoprosencephaly, microphthalmia, cardiac malformations, scalp defects, hypoplastic or absent ribs, and visceral and genital anomalies.

Along with all those phenotypic features, children with Down syndrome will be having congenital heart defects (50%), such as atrioventricular septal defects, ventricular septal defects, isolated secundum atrial septal defects, patent ductus arteriosus, and tetralogy of Fallot. Pulmonary involvement includes recurrent respiratory infections, sleep-disordered breathing, laryngo- and tracheobronchochomalacia, tracheal bronchus, pulmonary hypertension, and asthma. GI involvement includes congenital and acquired gastrointestinal anomalies as well as celiac disease. Other abnormalities include hypothyroidism, megak-aryoblastic leukemia, immune dysfunction, diabetes mellitus, seizures, alopecia, juvenile idiopathic arthritis, and problems with hearing and vision.(3)

Children with Down syndrome will experience developmental delays as well as limitations in everyday self-care activities such as dressing, personal hygiene, walking, and talking, as well as areas of health, education, and leisure. As a result, the demand on families of children with Down syndrome is substantially higher because these families must actively participate in their children's care. (1)

In India, the majority of people with mental retardation have traditionally been cared for by their families. The emotional and social stress that these parents undergo has been described by various investigators. Multifaceted factors have made these parents more vulnerable to stress.(2)

Developmental delay is universal in children with Down syndrome. However, cognitive impairment does not affect all areas of development in the same way. But all maladaptive behaviours in these people are inherently linked to cognitive impairment. Social development is relatively spared, but children with Down syndrome have difficulty using expressive language. Behaviour problems are relatively less in the majority of them. Psychiatric comorbidity is 18-38% in this population, which is lower than in children with similar levels of mental retardation from other aetiologies but still higher than in other children who are unaffected. Common behavioural difficulties include inattentiveness and stubbornness. All of these behaviours can respond to educational or pharmacologic interventions.(3)

If we intervene early through speech therapy, physiotherapy and occupational therapy and give proper medical attention for different health issues, children with Down syndrome will have a better long-term outcome as compared to other genetic causes of intellectual disability.(4)

More than 40 children attend our hospital's Down syndrome clinic to treat their health issues and co-morbidities. This aids in the early detection and appropriate treatment of their ailments. These therapies may increase their overall survival and long-term outcomes. Along with that, the psychological problems of their parents can also be addressed. The parents of these children have a lot of physical, mental, social, and economic problems while caring for their child with a disability. Western studies focused more on psychological problems among caregivers of Down syndrome children specifically. But Indian studies were mainly focused on caregivers of children with intellectual disabilities as a broad category, which also included Down syndrome.

Therefore, considering that we intend to conduct a study on the clinical and sociodemographic profile of children with Down syndrome and whether it is impacting the parents' everyday lives, social lives, and professional careers, is it a rising burden on their lives?

Method

A hospital-based descriptive study was conducted at the Regional Early Intervention Centre and the Department of Paediatrics, Government TD Medical College, Alappuzha. The study was conducted from November 2022 to July 2023 over a period of 9 months. Parental consent was taken at the time of direct interview of parents and children. The study included children with Down syndrome less than 18 years of age registered under the Regional Early Intervention Centre and the Department of Paediatrics, Government TD Medical College, Alappuzha. Children with Down syndrome who are registered under REIC and children who are getting admitted and consulting as outpatients at Government TD Medical College, Alappuzha, were included in our study. I explained the study and obtained consent for participation, as well as relevant information about the disease, therapy, and related investigations. A clinical examination was done, and caregiver burden was assessed using the Zarit Burden Interview scale.

According to a study by Neeraja Agarval Gupta, Diagnosis and Management of Down syndrome was published in October 2013. Prevalence of Down Syndrome is 0.125%.

Sample size was calculated using the formula

$$n = \frac{\left[Z^2_{1-\frac{\propto}{2}}\right](1-p)p}{\varepsilon^2 p}$$

P: expected proportion = 0.125

ε: Relative Precision = 5

1-∝/2: Desired confident level = 95

N: Population level = 40

The sample size adjusted for finite population is given by N_{finite}= f x n

Where, $F = \frac{1}{1 + \frac{n}{N_{Population}}}$

substituting all values sample size = 40

Data is obtained by personal interview using a standard proforma and consent from caregivers attending the Regional Early Intervention Centre and the Department of Paediatrics at Government T.D. Medical College, Alappuzha. These data will be transferred to Excel sheets. A structured proforma will be used to collect information from the patients. Subjects were interviewed by the questionnaire method, which included a burden interview, a sociodemographic profile, the Kuppuswamy scale and a clinical examination. The study will be initiated after getting approval from the IRC and the institutional ethics committee. After getting informed consent from the parents, data will be collected for a period of one year using a structured proforma. All the children registered under REIC Alappuzha and the Department of Paediatrics, Government Medical College, Alappuzha, are enrolled. These babies will be enrolled only after applying the exclusion criteria and getting consent from parents.

Statistical Analysis

Data will be tabulated using Microsoft Excel, then analysed and presented with the help of SPSS version 16. All qualitative variables are expressed as proportions. All quantitative variables are expressed as mean and standard deviation (SD). The association between variables will be studied using the chi-square test and the independent sample T-test.

Result

A total of 40 children with Down syndrome were included in this study. Out of the 40, 17 (42.5%) were female and 23 (57.5%) were male (Table 1 and Figure 1). The mean age of the children was 8.65 ± 4.65. Minimum age was 7 months and maximum age was 18 years. Out of 40 children in the <5-year age group, 6 (54.5%) were male and 5 (45.5%) were female. In 6-10 years, 6 (40.0%) were male and 9 (60%) were female. In 11-18 years, 5 (35.7%) were male and 9 (64.3%) were female. Out of 40 children, 20 (50.0%) of their mothers were aged 18-24, 15 (37.5%) were aged 25-34, and 5 (12.5%) were more than 35 years old (Table 2 and Figure 2). Among 40 children, in 16 (40%) cases, the education of the

father was graduation; in 9 (22.5%) cases, the education of the father was middle school certification; in 1 (2.5%) case, the education of the father was postgraduate; and in 14 (35%) cases, the education of the father was high school (Table 3 and Figure 3). Out of 40 children, in 8 (20.0%) cases, the education of the mother was graduation; in 28 (70.0%) cases, the education of the mother was high school; and in 4 (10.0%) cases, the education of the mother was middle school certification (Table 4 and Figure 4). Out of 40 children, the occupation of their father was found to be 5 (12.5%) in clerical, 6 (15.0%) in professional, 6 (15.0%) in semi-professional, 13 (32.5%) in semi-skilled, 1 (2.5%) in skilled owner and 9 (22.5%) in unskilled (Table 5 and Figure 5). Out of 40 children, the occupation of their mother was found to be 1 (2.5%) in clerical, 31 (77.5%) in homemaker, 1 (2.5%) in professional, 2 (5.0%) in semiskilled, 3 (7.5%) in skilled owner and 2 (5.0%) in unskilled (Table 6 and Figure 6). Out of 40 children, those included in the lower middle class were 4 (10%), in the upper SES were 2 (5%), in the upper lower were 21 (32.5%), and in the upper middle were 13 (32.5%) (Table 7 and Figure 7). Most of the families of children were from rural residences (23, 57.5%) and 17 (42.5%) from urban residences (Table 8 and Figure 8). Out of 40 children, 31 (77.5%) had their primary caregivers as mothers, 3 (7.5%) had their primary caregivers as fathers, and 6 (15%) had their primary caregivers as others (Table 9 and Figure 9). Out of 40 children, 22 children were in special school, 3 in normal school and 5 did not have schooling. In 10 cases, children aged less than 5 years were not going to school (Table 10 and Figure 10). Out of 40 children, more than half of the children had a mild to moderate burden. 24 (60%) and 16 (40%) had little or no burden (Table 11 and Figure 11). Among these children, the weight of most of them was 23 (57.5%) between the 25th and 75th centiles, 4 (10%) below the 5th centile, 12 (30%) between the 5th and 25th centiles, no child between the 75th and 95th centiles and 1 (2.5%) above the 95th centile (Table 12 and Figure 12). Children with a weight between the 25th and 75th centiles were in the normal range. The 75th to 95th was high normal, and above the 95th centile was overweight. Children with a weight between the 5th and 25th centiles were low normal, and those below the 5th centile were underweight. Among these children, the height of most of them was 24 (60.0%) between the 25th and 75th centiles, 3 (7.5%) below the 5th centile, 7 (7.5%) between the 5th and 25th centiles, 5 (12.5%) between the 75th and 95th centiles, and 1 (2.5%) above the 95th centile (Table 13 and Figure 13). Children with a height between the 25th and 75th centiles were in the normal range. The 75th to 95th was high normal, and above the 95th centile was tall. Children with a height between the 5th and 25th centiles were low normal, and those below the 5th centile were of short stature. Among these children, the head circumference of most of them is 28 (70.0%) between the 25th and 75th centiles, 4 (10.0%) below the 5th centile, 2 (5%) between the 5th and 25th centiles, 5 (12.5%) between the 75th and 95th centiles, and 1 (2.5%) above the 95th centile (Table 14 and Figure 14). The most common heart disease in children with Down syndrome among our study population was Ostium Secundum ASD, followed by bicuspid aortic valve and VSD (Table 15 and Figure 15). Then there were Ostium Primum ASD, Ostium Secundum ASD, PDA, Ostium Secundum ASD with PDA and TOF, one case each. Out of 40 children, 4 (10.0%) had seizures, 1 (2.5%) had contractures, 1 (2.5%) had hypertonia, 39 (97.5%) had hypotonia, and 7 (17.5%) had speech abnormalities requiring speech therapy (Table 16 and Figure 16). Out of 40 children, 1 (2.5%) had moderate ID, 17 (42.5%) had mild ID and 11 (27.5%) had borderline ID (Table 17 and Figure 17). The remaining 11 children are less than five years old. So the Developmental Quotient was completed. All of them had a value less than 70, with a mean of 63.63, indicating developmental delay. 20 (50.0%) of children had hypothyroidism, and 20 (50%) had normal thyroid function (Table 18 and Figure 18). Out of 40 children, 17 (42.5%) were anaemic (Table 19 and Figure 19).

Table 1 Distribution of Age and Sex

Age group	Sex			
	Male		Female	
	No	%	No	%
<5 year	6	54.5	5	45.5
6-10 year	6	40	9	60
11-18 year	5	35.7	9	64.3

Figure 1

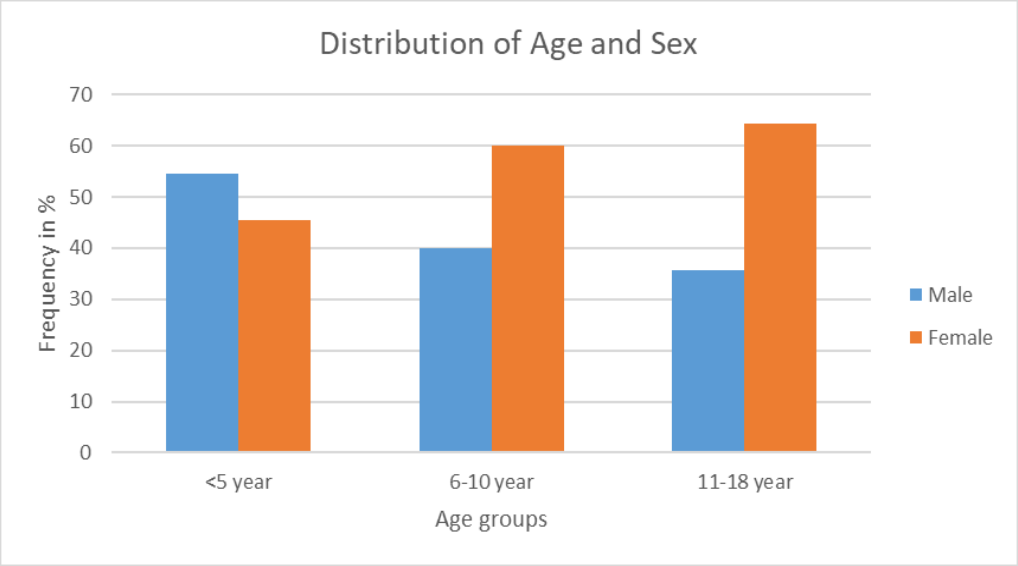


Table 2 Distribution Maternal Age at Pregnancy

Maternal Age	No.	%
18-24	20	50.0
25-34	15	37.5
>=35	5	12.5

Figure 2

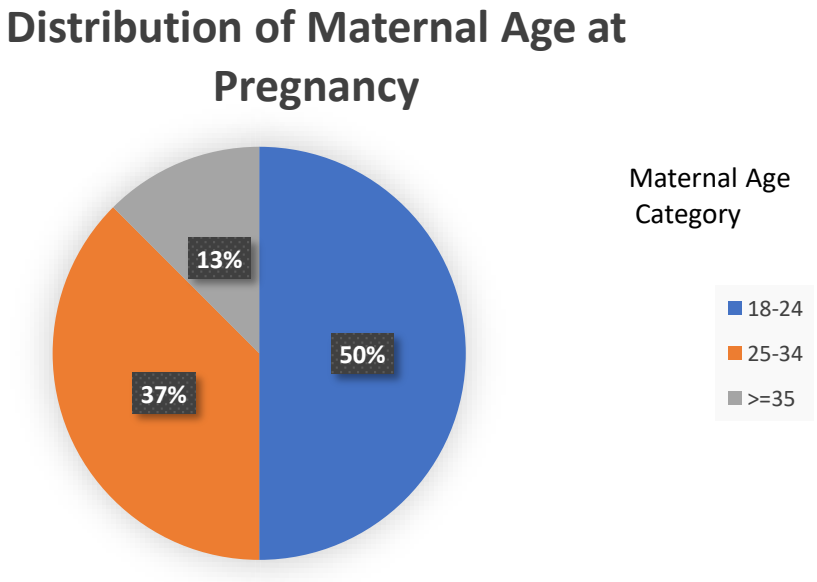


Table 3 Distribution of Education of Father

Education of father	No.	%
Post Gradate	1	2.5
Graduate	16	40.0
High School	14	35.0
Middle School Certification	9	22.5
Total	40	100.0

Figure 3

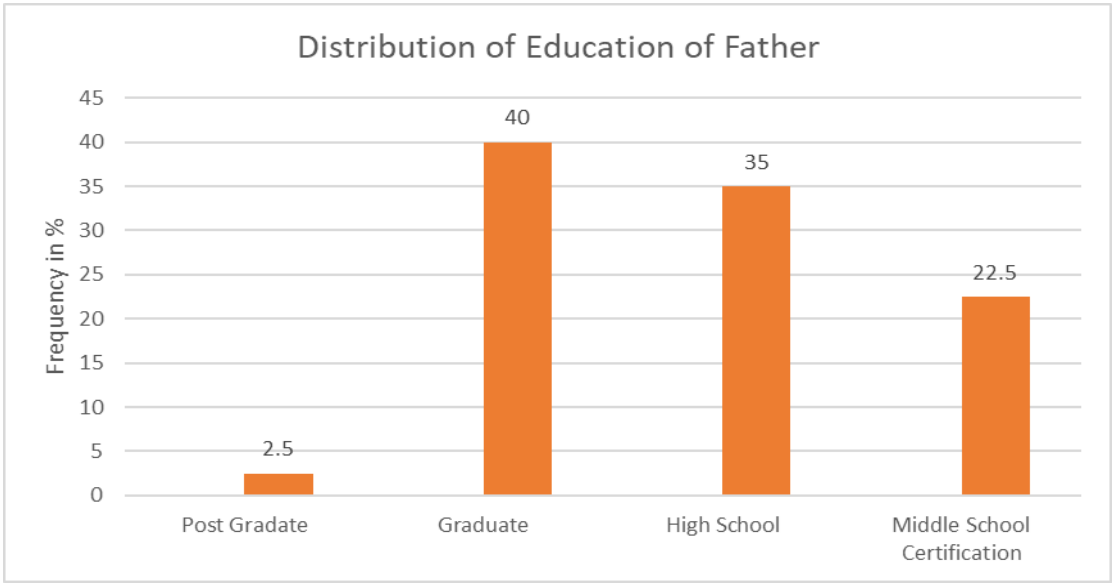


Table 4 Distribution of Education of Mother

Education of Mother	No.	%
Graduate	8	20.0
High School	28	70.0
Middle school certification	4	10.00
Total	40	100.0

Figure 4

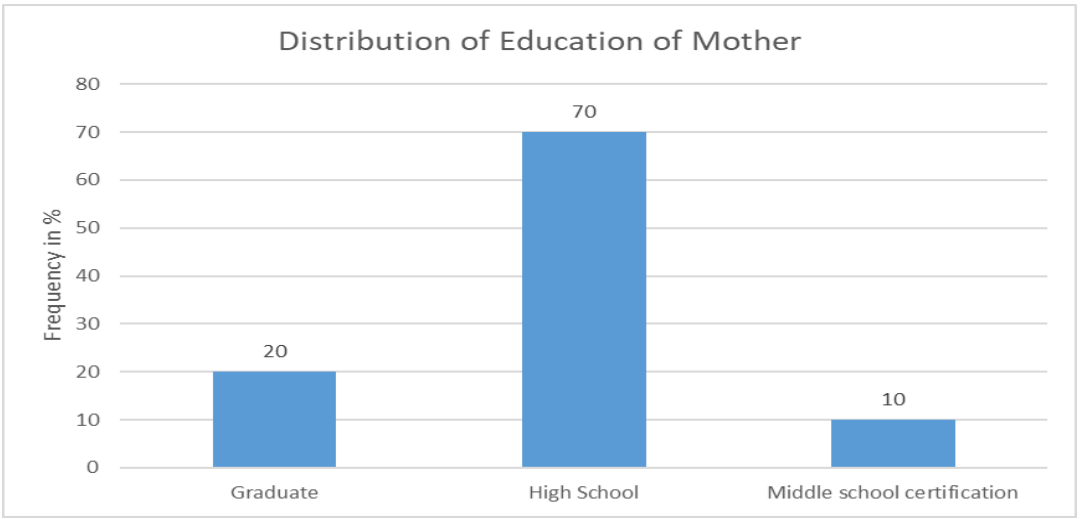


Table 5 Distribution of Occupation Father

Occupation father	No	%
Profession	6	15.0
Semi Profession	6	15.0
Clerical	5	12.5
Skilled Owner	1	2.5
Semiskilled	13	32.5
Unskilled	9	22.5

Figure 5

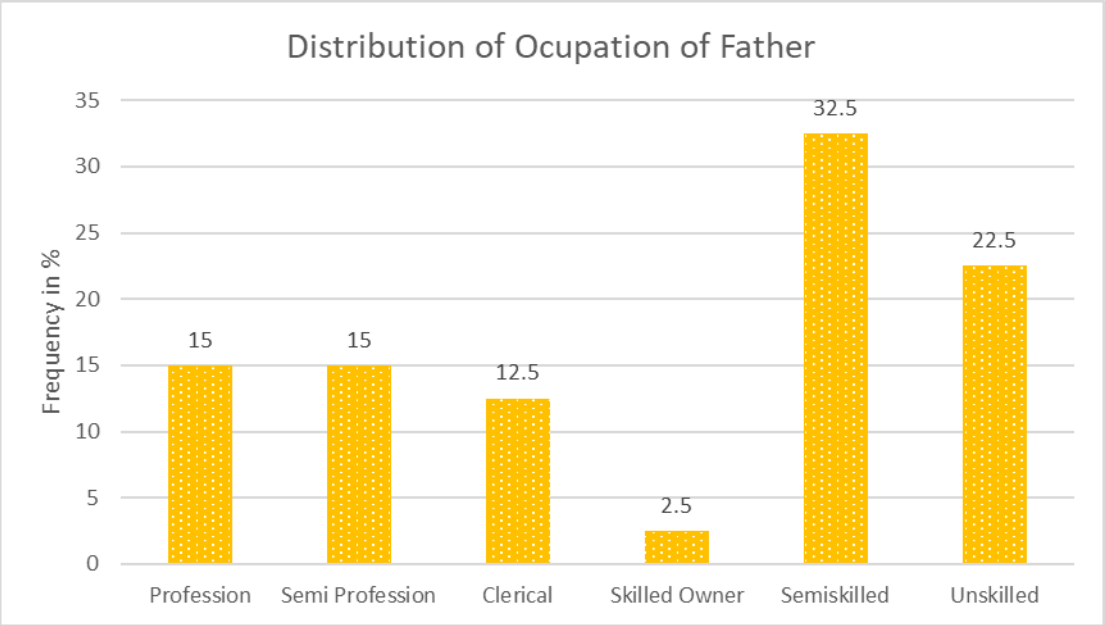


Table 6 Distribution of Occupation of Mother

Occupation of Mother	Number	%
Profession	1	2.5
Clerical	1	2.5
Skilled owner	3	7.5
Semiskilled	2	5.0
Unskilled	2	5.0
Home maker	31	77.5

Figure 6

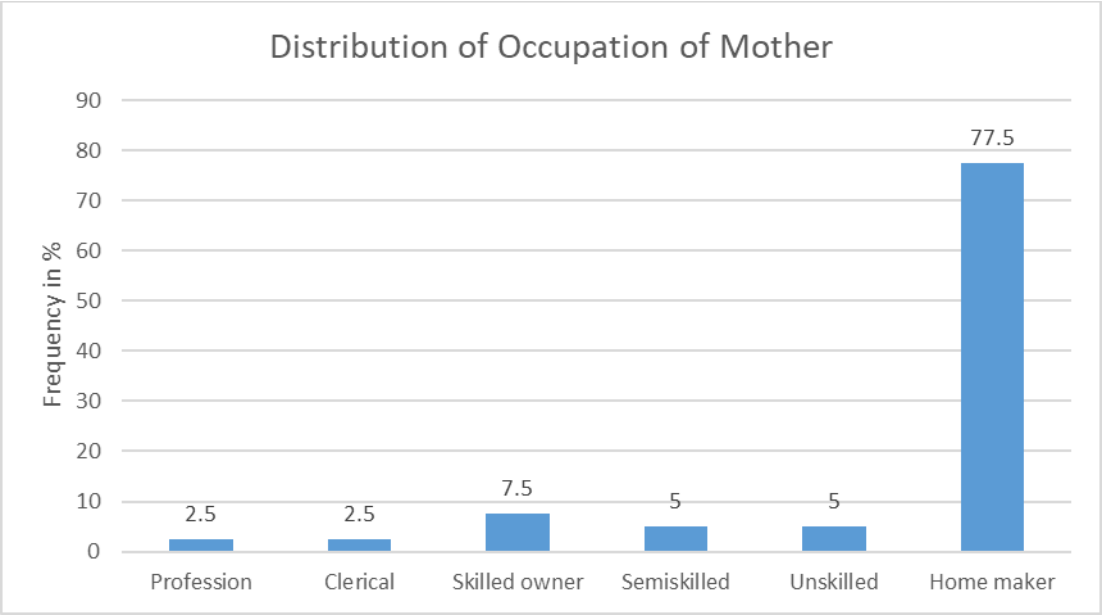


Table 7 Distribution of Socio-Economic Status

Socio Economic Status	No.	%
Upper	2	5.0
Upper Middle	13	32.5
Lower Middle	4	10.0
Upper Lower	21	52.5
Total	40	100.0

Figure 7

Table 8 Area of Residence

Residence	No.	%
Rural	23	57.5
Urban	17	42.5
Total	40	100.0

Figure 8

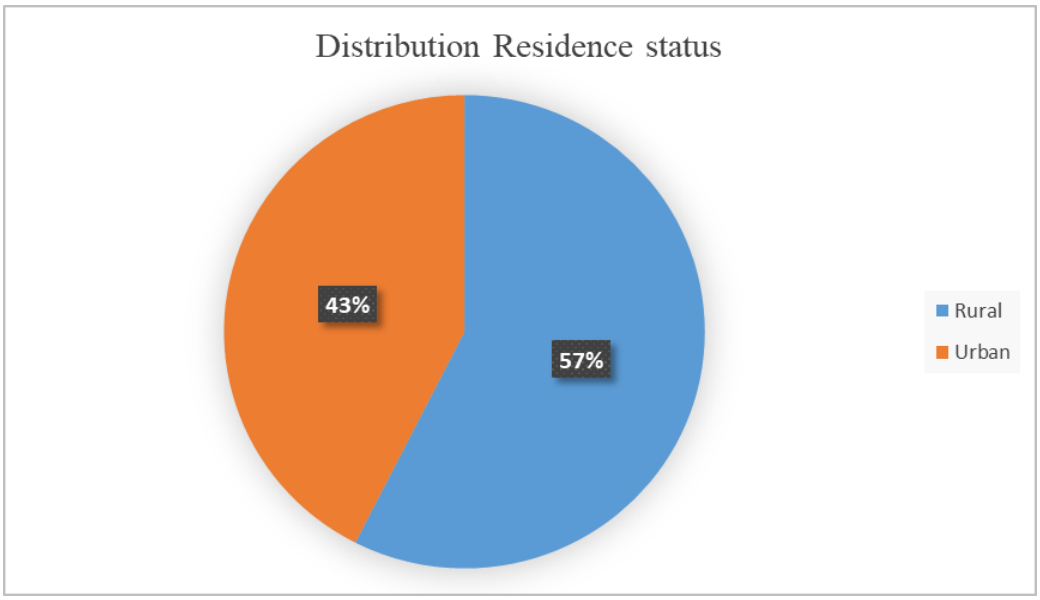


Table 9 Primary Caregiver of the Child

Caregivers Status	No.	%
Mother	31	77.5
Father	3	7.5
Others	6	15.0
Total	40	100.0

Figure 9

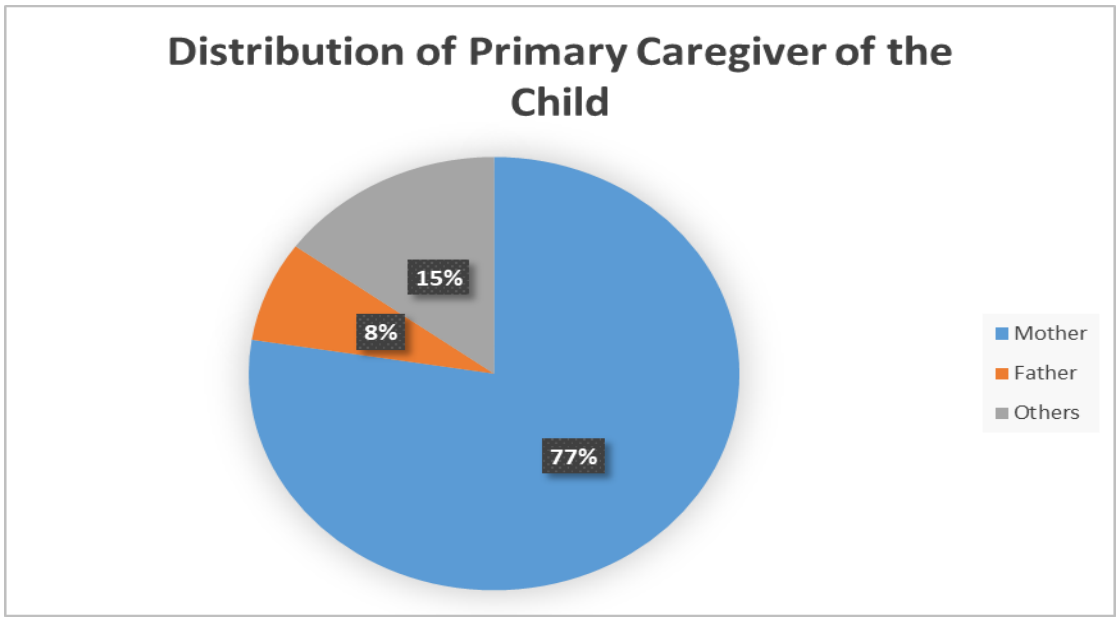


Table 10 Distribution of Schooling

Schooling	No.	%
Not Applicable	10	25.0
No Schooling	5	12.5
Normal School	3	7.5
Special School	22	55.0
Total	40	100.0

Figure 10

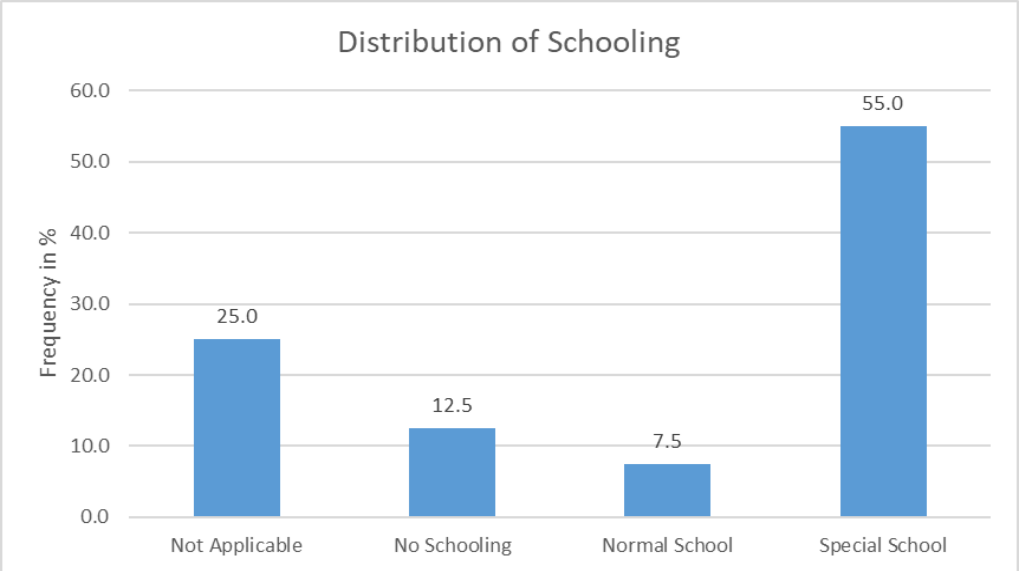


Table 11 Distribution of Caregiver Burden

Burden	No.	%
Little or No Burden	16	40.0
Mild to Moderate Burden	24	60.0
Total	40	100.0

Figure 11

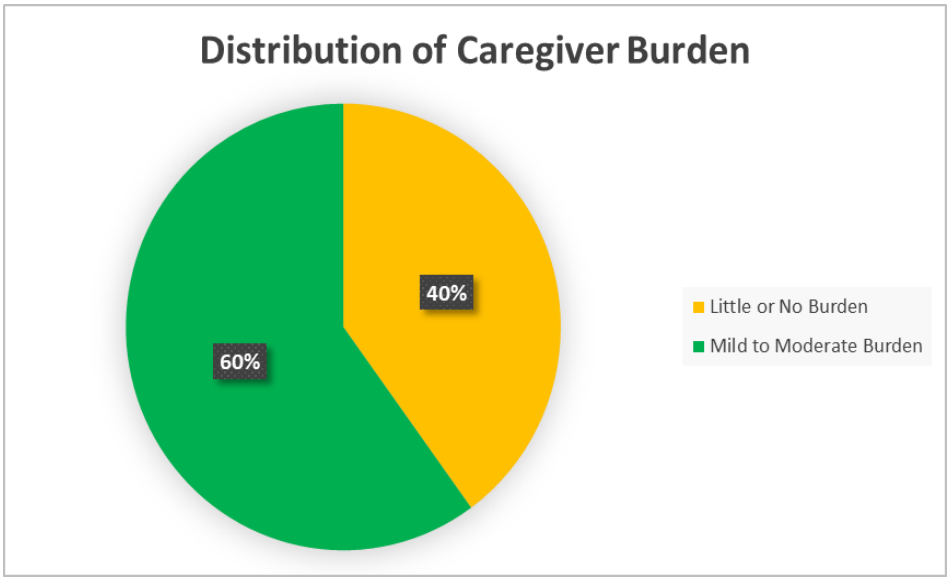


Table 12 Distribution of Weight

Weight	No.	%
Below 5th centile	4	10
5th to 25th centile	12	30
25th to 75th centile	23	57.5
75th to 95th centile	0	0
Above 95th centile	1	2.5

Figure 12

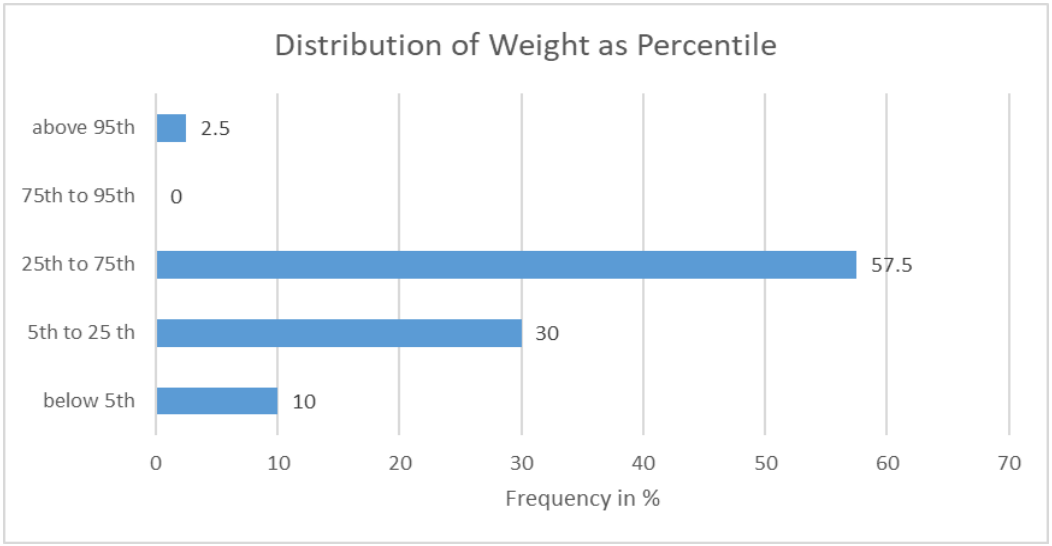


Table 13 Distribution of Height

Height	Number	%
below 5th	3	7.5
5th to 25 th	7	17.5
25th to 75th	24	60
75th to 95th	5	12.5
above 95th	1	2.5

Figure 13

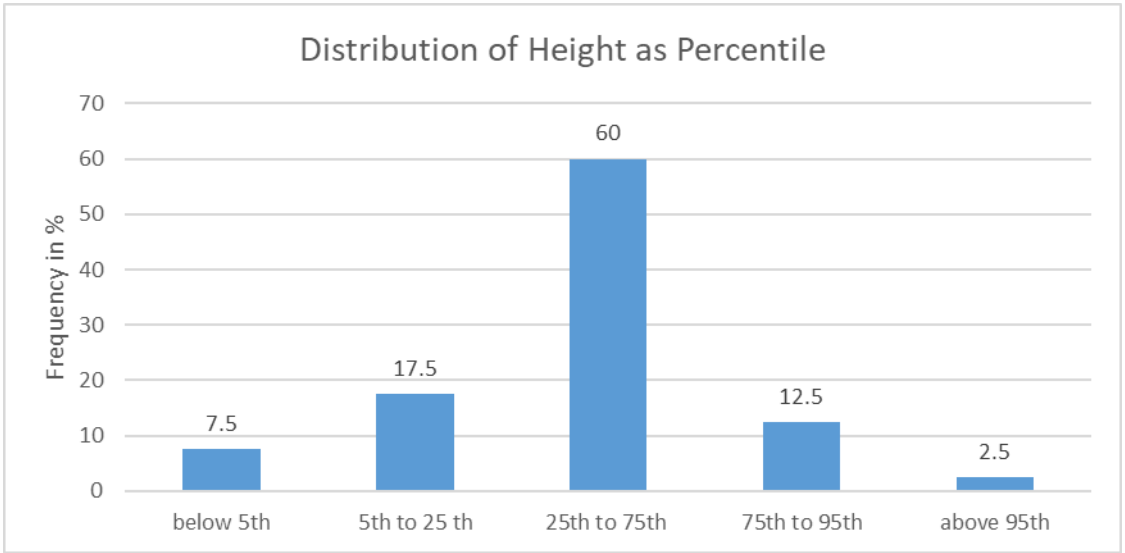


Table 14 Distribution of Head Circumference

Head circumference	No.	%
below 5th	4	10
5th to 25 th	2	5
25th to 75th centile	28	70
75th to 95th	5	12.5
above 95th	1	2.5

Figure 14

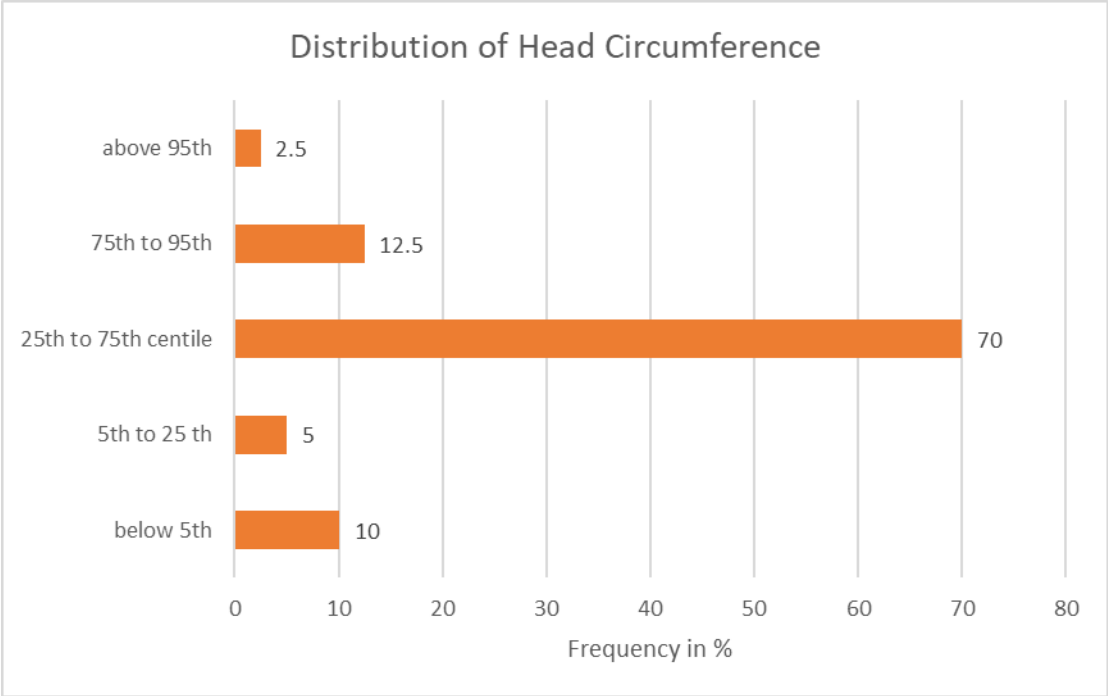


Table 15 Congenital Heart Disease

Congenital Heart Disease	No.	%
Bicuspid Aortic Valve	3	7.5
OP ASD	1	2.5
OS ASD	8	20.0
OS ASD PDA	1	2.5
PDA	1	2.5
TOF	1	2.5
VSD	3	7.5

Figure 15

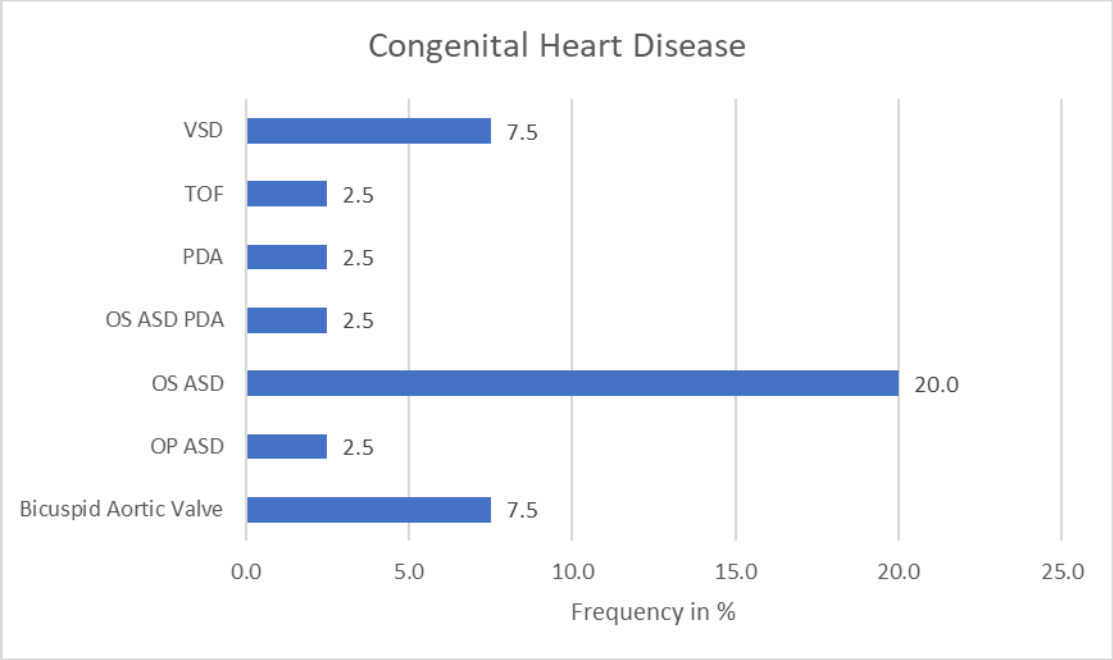


Table 16 Distribution of Neurological features

Neurological	No.	%
Seizures	4	10.0
Contractures	1	2.5
Hypertonia	1	2.5
Hypotonia	39	97.5
Speech Abnormalities requiring speech therapy	7	17.5

Figure 16

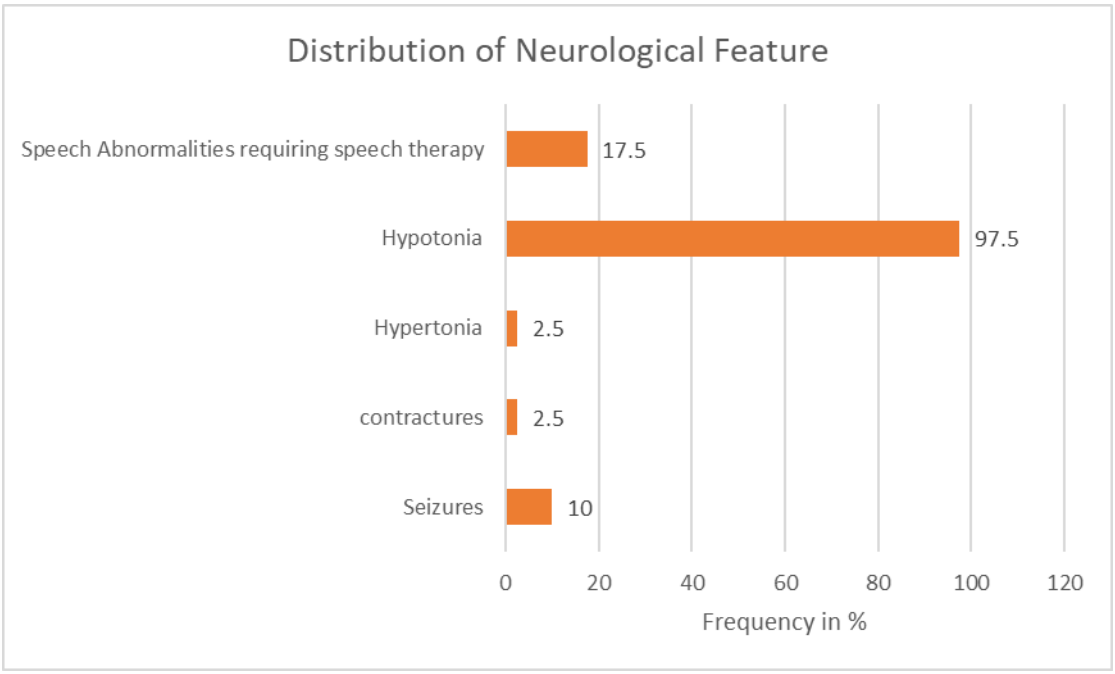


Table 17 Distribution of IQ Score

IQ Score	No.	%
Moderate ID	1	2.5
Mild ID	17	42.5
Borderline	11	27.5
Total	29	72.5

Figure 17

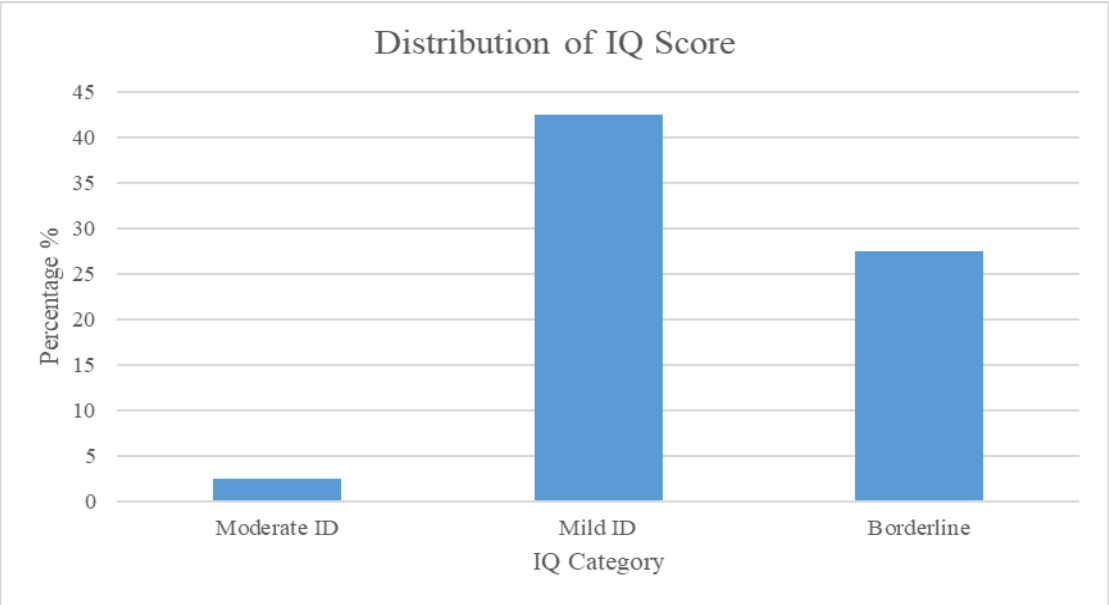


Table 18 Thyroid Functional Status

Thyroid Status	No.	%
Hypothyroidism	20	50.0
Normal	20	50.0
Total	40	100.0

Figure 18

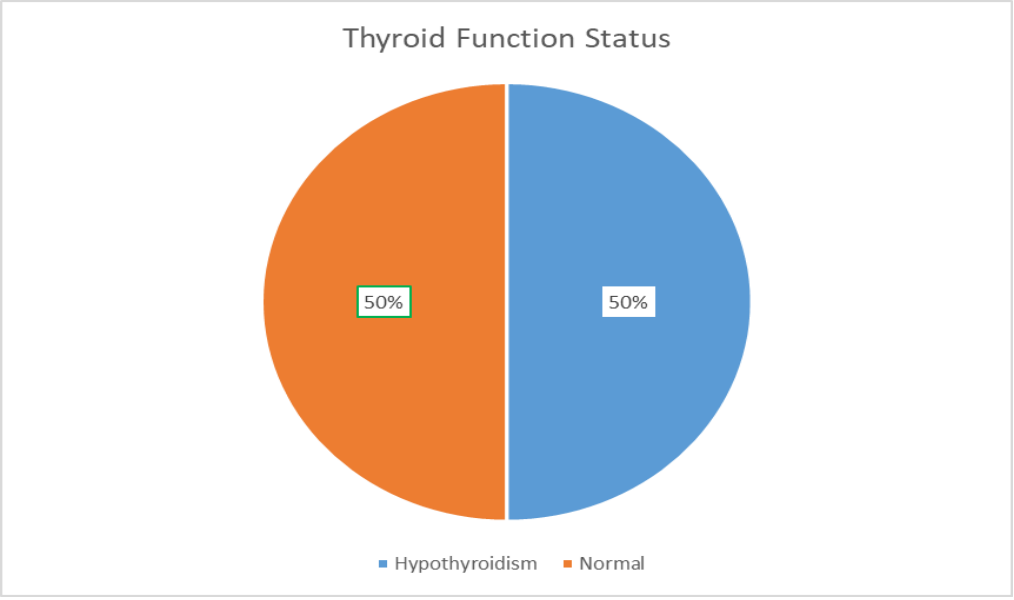
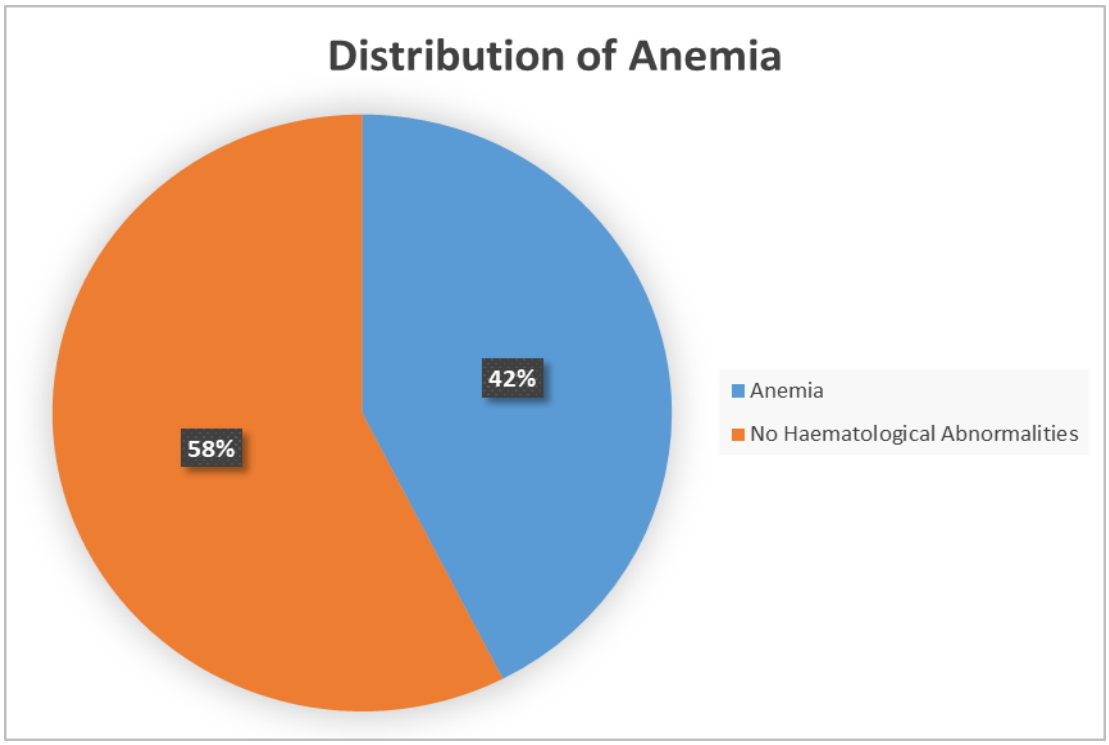


Table 19 Distribution of Anemia

Haematological Abnormalities	No.	%
Anemia	17	42.5
No Haematological Abnormalities	23	57.5

Figure 19



Discussion

The study "Sociodemographic and Clinical Profile of Down Syndrome Children Less Than 18 Years and Its Effect on Caregiver Burden in a Tertiary Care Centre" was conducted at Govt. T.D. Medical College, Alappuzha, and included 40 cases of children with Down syndrome. In our study population, there are more men than women. In a study conducted by Verma R S and Haq A, the male-to-female ratio was 1.3, whereas in our study it was 1.35. Out of 40 children in our study, the majority of their mothers were between the ages of 18 and 24. Even though the chance of having a kid with Down syndrome increases with age, since the reproductive age group consists of younger women, mothers of younger ages have a greater incidence of Down syndrome babies. Other studies have produced similar results.[6] The majority of children with Down syndrome in our study population were between the ages of 6 and 10. Among the 40 children, the majority had fathers who had completed high school, middle school certification, and postgraduate studies. Among 40 children, the majority of them had mothers with high school education, followed by graduation and middle school certification. The majority of our children fall into the upper lower and upper middle socioeconomic strata. There is research that shows people with Down syndrome have unmet needs and have a greater impact on their families. These differences become more pronounced for children without insurance and from low socioeconomic status families. (76) Financial burden is a common phenomenon, often noticed in the caregivers of children with Down syndrome. It echoes adverse effects on the caregiver's mental and physical health. [8] Financial burden is a common phenomenon, often noticed in the caregivers of children with Down syndrome. It echoes adverse effects on the caregiver's mental and physical health.[7]

Most of the families of children in our study population were from rural residences. The majority of our children had a mother as their primary caregiver. Other studies also point out that mothers are the usual caregivers of Down syndrome children. [5] Most of the children in our study population are attending special schools. Height, weight, and head circumference were measured in all children among our study population. Among all the measurements of the study population, the majority of observations remain between the 25th and 75th centiles. The most common heart disease in children with Down syndrome among our study population was Ostium Secundum ASD, followed by bicuspid aortic valve and VSD. Caregiver burden among children with congenital heart disease was statistically significant in our study population. Other studies also cited similar findings. review of recent advances and clinical implications. Jun Yasuhara et al. pointed out in their studies that advances in clinical management have resulted in tremendous improvement in the survival of children with CHD. But adult survivors have to encounter cardiac and non-cardiac comorbidities, which affect quality of life and prognosis.[9]

Neurological evaluation showed the child having a seizure, contracture, hypotonia, hypertonia, and speech abnormality. All of the children exhibited speech abnormalities and hypotonia. Seven children with speech problems are undergoing speech therapy. All children under the age of five had a Developmental Quotient (DQ) of less than 70. The Intelligence Quotient (IQ) was below 80, with a maximum value of 68. Out of 29 children aged more than 5 years, 18 were mentally retarded and 11 were borderline. Down syndrome (DS) is associated with variable intellectual disability and multiple health and psychiatric comorbidities. These comorbidities have an effect on the cognitive outcomes of the child, which in turn affects quality of life. Anaemia was identified as the most prevalent haematological issue in our study. Half of the participants in our research population were diagnosed with hypothyroidism and were undergoing treatment for this condition. Refractory error was the most common ocular problem, followed by squint. But these problems don't cause any increased caregiver burden among parents. Hearing was normal among all our children in the study population. Dental caries is the most common dental problem among our study population, followed by malalignment. One child got fluorosis. Most of these findings are overlapping among our study population. Among GIT problems, 15 of our study population got constipation. No other GIT-related symptoms are there in our study population. None of them had any surgical problems. Shoulder dislocation is the most common skeletal abnormality seen in our study population. Since no one among our study population had any symptoms and none of them are planning to engage in any contact sports, atlantoaxial instability was not screened.

Conclusion

There are more males than females in our research population. The maternal age group is among the youngest during pregnancy. In our study population, fathers have greater educational and occupational levels than mothers. The socioeconomic majority of them are from the lower class. The study population is primarily composed of rural residents. The mother is the primary caregiver for most of the children in our study. The majority of the children attend special school. A mild-moderate class of modified Zarit's caregiver burden is common among our study population. Anthropometry indicated that the majority of the study population had normal height, weight, and head circumference. Hypothyroidism is present among 50% in our study population. Mild intellectual disability is more common among our study population. Hypotonia is present among most of our children. OSASD is the most common congenital heart disease among our study population.

Statements and Declarations

This manuscript represents original research conducted by the authors, and it has not been published or submitted elsewhere for consideration. We affirm that all authors have contributed significantly to this work and have approved the final

version of the manuscript. There are no conflicts of interest to disclose.

Reference

1. Agarwal Gupta N, Kabra M. Diagnosis and management of Down syndrome. Indian J Pediatr. 2014 Jun;81(6):560-7. doi: 10.1007/s12098-013-1249-7. PMID: 24127006.
2. Azeem MW, Dogar IA, Shah S, Cheema MA, Asmat A, Akbar M, Kousar S, Haider II. Anxiety and depression among parents of children with intellectual disability in Pakistan. J Can Acad Child Adolesc Psychiatry. 2013 Nov;22(4):290-5. PMID: 24223048; PMCID: PMC3825469.
3. Kliegman R, Geme JWS. Nelson Textbook of Pediatrics [Internet]. 21st ed. Elsevier; 2019. Available from: https://books.google.co.in/books?id=k85owAEACAAJ
4. Buckley S. Living with Down syndrome. 2000 Jan 1. Down Syndrome Issues and Information. Issues for families - Down syndrome [Internet]. 2014 [cited 2014 Oct 20]. Available from: http://www.downsyndrome.org/information/family
5. Hodapp RM. Parenting children with mental retardation. In: Bornstein MH, editor. Handbook of parenting. 2nd ed. Vol. 1. Mahwah, NJ: Lawrence Erlbaum; 2002. p. 355-82.
6. MacLennan S. Down's syndrome. InnovAiT. 2020;13(1):47-52. doi: 10.1177/1755738019886612.
7. Shetty J, Shetty A, Mundkur SC, Dinesh TK, Pundir P. Economic burden on caregivers or parents with Down syndrome children—a systematic review protocol. Syst Rev. 2023 Jan 6;12(1):3.
8. McGrath RJ, Stransky ML, Cooley WC, Moeschler JB. National profile of children with Down syndrome: disease burden, access to care, and family impact. J Pediatr. 2011 Oct;159(4):535-40.e2. doi: 10.1016/j.jpeds.2011.04.019. Epub 2011 Jun 12. PMID: 21658713.
9. Yasuhara J, Garg V. Genetics of congenital heart disease: a narrative review of recent advances and clinical implications. Transl Pediatr. 2021 Sep;10(9):2366-2386. doi: 10.21037/tp-21-297. PMID: 34733677; PMCID: PMC8506053.
10. Startin CM, D'Souza H, Ball G, Hamburg S, Hithersay R, Hughes KMO, et al. Health comorbidities and cognitive abilities across the lifespan in Down syndrome. J Neurodev Disord. 2020 Jan 23;12(1):4.