

Original Research Article

Medical problems in children with Down syndrome

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ABSTRACT

Background: Down syndrome (DS) is a major cause of mental retardation of prenatal origin and has several associated co-morbidities involving cardiovascular system, respiratory, endocrine system, hematological, gastrointestinal, musculoskeletal, eye and ear defects, immunological changes and neurological system. This study was conducted to identify the common medical problems in children with Down syndrome and the morbidity associated with these conditions. The objective of the present study was to find out the occurrence of different medical problems in children with DS.

Methods: 42 children with a phenotype of Down syndrome in the age group of 0-12 years attending the outpatient, inpatient and Down syndrome Clinic of the Institute of Child Health, Kottayam during the study period were included in the study by purposive sampling. Demographic details were entered, and Pediatric Clinical Examination was performed by the investigator himself to identify the medical problems. Old medical reports were reviewed, and data entered into a proforma and statistically analysed.

Results: Out of the 42 children with DS, 22 were males. 15 (35.7%) were less than 1 year, 20 (48.3%) children 1-5 years and 7 (16.1%) children 5-12 years of age. Mean age of the study group was 1.78 ± 0.51 years. Mean age of their mothers at the time of conception was 30.6 ± 5.8 years. 26 (57%) children with Down syndrome had a medical problem during the neonatal period which required hospitalization. Almost all systems are affected and craniofacial features, developmental delay and hypotonia were universal. Various forms of congenital heart diseases were observed in 67% and hypothyroidism in 23.8%.

Conclusions: Down syndrome is a common genetic disorder with multisystem involvement. Congenital heart diseases, hypothyroidism and recurrent respiratory infections were the common medical problems identified in this study.

Keywords: Comorbidities, Down syndrome, Medical problems

INTRODUCTION

Down syndrome (DS) is the commonest genetic disorder in children with an incidence of 1 in 700-800 live births. Down Syndrome is a major cause of mental retardation of prenatal origin and has several associated comorbidities involving cardiovascular system, respiratory, endocrine system, hematological, gastrointestinal, musculoskeletal,

eye and ear defects, immunological changes and neurological system.¹

Recent studies show average age of mother giving birth to Down syndrome child around 26-27 years age, may be due to early completion of reproductive cycle. Most of the anomalies in DS children can be detected in the antenatal period itself, so that parents can be informed and anticipate the difficulties, and make necessary

arrangements if needed, after birth of the child. Developmental delay is universal. Cognitive impairment doesn't uniformly involve all areas of development. Social development is relatively spared, but children with DS have difficulty in using expressive language. The majority of children with DS don't have behavior problems, if present most of them are secondary to cognitive impairment.

The child with DS poses a significant challenge to the family and rearing these children to the optimum level is still more challenging. The birth of a DS child often contributes much stress and anxiety to the parents. When compared to normal children these babies require frequent medical attention and supportive care. Proper medical care and parental education can improve their quality of life and life skills.²

Hence this study was conducted to identify the common medical problems in children with Down syndrome and the morbidity associated with these conditions.

METHODS

Forty-five children with a phenotype of Down syndrome in the age group of 0-12 years attending the outpatient, inpatient and Down syndrome Clinic of the Institute of Child Health, Kottayam during the study period July 2016 to January 2017 were consecutively included in the study by purposive sampling.

The purpose and details of the study, ethical principles adopted, and confidentiality of the data handling were explained to the parents and informed consent for participation was obtained. Three of them refused participation and were excluded and finally 42 children got included. Data was collected after getting the Institutional Review Board approval and permission from hospital authorities. Demographic details were entered into a general basic data sheet. Focused history of medical illness and surgical problems from birth onwards were collected and verified with the old records whenever available. Pediatric Clinical examination was performed by the investigator himself to identify the present medical condition and nutritional status and data entered into a proforma.

Development screening was done for all, though formal IQ assessment was not done. Growth and nutritional status assessed using Indian Academy of Pediatrics (IAP) growth charts and graded according to the IAP Grading of Protein-Energy Malnutrition (1972).³ Data was analyzed using appropriate descriptive statistics as measures of central tendency viz. percentage, mean and standard deviation.

RESULTS

Out of the 42 children with DS, 22 were males and 20 females. 15 (35.7%) were less than 1 year, 20 (48.3%)

children 1-5 years and 7 (16.1%) children 5-12 years of age. Mean age of the study group was 1.78 ± 0.51 years. Most of the mothers were below 40 years of age. 38.7% belonged to the age category 35-40 years.

Contrary to the belief that children with DS are born to elderly mothers, 16 (38.1%) were below 30 years of age at the time of conception of their DS baby. Mean age at conception was 30.6 ± 5.8 years.

Table 1: Neonatal problems in the study group.

| NICU admission | Frequency | Percentage |
|----------------------|-----------|------------|
| Hyperbilirubinemia | 9 | 21.4 |
| Respiratory distress | 6 | 14.4 |
| Feeding difficulty | 9 | 21.4 |
| No NICU admission | 18 | 42.8 |
| Total | 42 | 100 |

This study showed various medical problems in children with Down syndrome. Craniofacial features which are phenotypical peculiarities of DS were found in all cases in varying degrees. These included hypertelorism, epicanthic folds, depressed nasal bridge, low set ears, brachycephaly etc.

Table 2: Common medical problems observed in the study.

| Medical problems | Frequency (N=42) | Percentage |
|---------------------------|------------------|------------|
| Craniofacial dysmorphism | 42 | 100 |
| Cardiac | 28 | 66.7 |
| Respiratory | 24 | 57.1 |
| CNS (Hypotonia) | 41 | 97.6 |
| Developmental delay | 42 | 100 |
| Hypothyroidism | 10 | 23.8 |
| Urological | 1 | 2.3 |
| Eyes (Strabismus) | 27 | 64.2 |
| Hematology | | |
| Anemia | 4 | 9.5 |
| Acute Leukemia | 2 | 4.7 |
| Clinodactyly | 24 | 57.1 |
| Atlanto-axial subluxation | 2 | 4.7 |
| Microcephaly | 27 | 65.0 |

26 out of 42 (57%) children with Down syndrome had a medical problem during the neonatal period which required hospitalization. Neonatal hyperbilirubinemia (jaundice) and feeding difficulties were the common reasons for admission in this series.

Almost all systems are affected and craniofacial features which are characteristic of the condition were present in all children. Developmental delay and hypotonia were also universal. Various forms of congenital heart diseases (CHD) were observed in almost two thirds (67%).

Table 3: Cardiac lesions noted in children with DS.

| Cardiac lesion | Frequency | Percentage |
|---------------------------------|-----------|------------|
| Ventricular septal defect (VSD) | 8 | 19.0 |
| Atrial septal defect (ASD) | 14 | 33.3 |
| Patent ductus arteriosus (PDA) | 1 | 2.3 |
| AV canal defect (AVCD) | 4 | 9.5 |
| Tetralogy of fallot (TOF) | 2 | 4.7 |
| No cardiac anomaly | 14 | 33.3 |
| Total | 42 | 100 |

Table 3 shows the distribution of different congenital heart diseases in children with DS in the present series. ASD was the commonest anomaly in this series. Cardiac anomalies were observed in two thirds of the children. Seven underwent surgical correction and the remaining are under medical follow up (Table 4).

Table 4: Surgical interventions done.

| Surgery | Frequency | Percentage |
|---------------------------|-----------|------------|
| Duodenal atresia | 3 | 7.1 |
| Pyeloplasty | 1 | 2.3 |
| TOF (intracardiac repair) | 1 | 2.3 |
| VSD/AV canal repair | 6 | 14.2 |
| Cervical spine fixation | 2 | 4.6 |
| No surgery | 29 | 69.0 |
| Total | 42 | 100 |

11 out of 42 (26.2%) children required a major surgical intervention. 16.5% underwent cardiac surgeries. Three underwent laparotomies for duodenal atresia.

Table 5: Nutritional status of children with Down syndrome.

| Grade | Frequency | Percentage |
|-------------------------|-----------|------------|
| No PEM (wt >80 %) | 24 | 57.1 |
| Grade 1 PEM (wt 71-80%) | 10 | 24.8 |
| Grade 2 PEM (wt 61-70%) | 4 | 9.5 |
| Grade 3 PEM (wt 51-60%) | 4 | 9.5 |
| Grade 4 PEM (wt ≤50%) | 0 | 0 |
| Total | 42 | 100 |

Table 5 shows the distribution of protein energy malnutrition in the study group. More than 50% of the DS children in this series did not show malnutrition based on the weight for age. Only 9.5% had severe degree of malnutrition in the present study according to the IAP classification 1972. 61.3% had normal height for age according to Waterlow's classification.⁴ Severe stunting was noted only in 9.5 %.

DISCUSSION

Majority of the children 35 (84.0%) were below 5 years of age and the mean age of the study group was

1.78±0.51 years. This is due to the fact that this is a hospital-based study and the medical problems' which require frequent hospital care are more prevalent in the younger age group. The medical follow up is frequent and compliance to medical advices is more during the early years of life. As the age increases the health status stabilizes and the need of frequent hospital visits decline.

Most of the mothers were below 40 years of age. 38.7% belonged to the age category 35-40 years. Contrary to the belief that children with DS are born to elderly mothers, 16 (38.7%) were below 30 years of age at the time of conception of their DS baby. Mean age at conception was 30.6±5.8 years.

Various medical problems were identified in these children. 58% children required NICU admissions during the newborn period. 9 (21.4%) had hyperbilirubinemia, 6 (14.4%) had respiratory distress, and 9 (21.4%) had feeding difficulties. Craniofacial features which are phenotypical peculiarities of DS were found in all cases in varying degrees. Brachycephaly, microcephaly, a sloping forehead, a flat occiput, large fontanels with late closure, a patent metopic suture, absent frontal and sphenoid sinuses, and hypoplasia of the maxillary sinuses occur. Up-slanting palpebral fissures, bilateral medial epicanthic folds, Brushfield spots (speckled iris), refractive errors (50%), strabismus (44%), nystagmus (20%), blepharitis (33%), conjunctivitis, tearing from stenotic nasolacrimal ducts, congenital cataracts (3%), pseudo papilledema, spasm nutans, acquired lens opacity (30-60%), retinal detachment, and keratoconus in adults are observed.⁵

Developmental delay and hypotonia were universal. 28 (66.7%) had Congenital heart disease, 10 (23.8%) had hypothyroidism and 20 (64.5%) had strabismus (squint). Commonest CHD identified in this study was ostium secundum ASD. Mihci E et al, reported 72.3% prevalence of CHD in their study and the commonest lesion was ostium secundum ASD (25%).⁶ Kava MP et al, reported 18.3% prevalence of CHD in their study and Kapoor S et al, reported a higher prevalence of 45%.^{7,8}

Moderate-to-severe intellectual disability occurs as a constant feature, with IQs ranging from 20 to 85 (mean, approximately 50). Muscle hypotonia is seen in newborns with decreased response to normal stimuli; this improves with age. Seizure disorders are present in 5-10% or patients.⁹ Infantile spasms are the most common seizures observed in infancy, whereas tonic-clonic seizures are most common in older patients. However, seizures were not reported in this study.

Two (4.7%) children developed atlanto-axial subluxation which was treated surgically. Toddlers and school-aged children with DS should be screened for atlanto-axial subluxation using lateral radiographs of the neck in flexion and extension. Pueschel SM et al, observed that approximately 10% to 20% of children have atlanto-axial

subluxation, while 3% display cervical myelopathy. The average age of onset of myelopathy is 10.5 years in that series.¹⁰ Surgical stabilization is required to avoid quadriplegia if instability is noted and there are signs of spinal cord compression. The need for screening and periodic medical checkups for these children cannot be over emphasized for early detection and prevention of such complications.

GI abnormalities occur in approximately 12% of patients.¹¹ Duodenal atresia or stenosis, Hirschsprung disease, tracheoesophageal fistula, Meckel diverticulum, imperforate anus, and omphalocele are observed. Gastroesophageal reflux is commonly seen in children with Down syndrome and can be severe enough to result in aspiration of stomach contents, resulting in respiratory symptoms such as persistent coughing, wheezing, and pneumonia. Infants with oral-motor difficulties may present with choking and gagging on feedings as well as the respiratory symptoms mentioned. Three children in this study had duodenal atresia which was operated.

The prevalence of thyroid disorders (e.g., congenital hypothyroidism, primary hypothyroidism, autoimmune thyroiditis, and compensated hypothyroidism or hyperthyrotropinemia) is reportedly 3-54% in individuals with Down syndrome and increases with increasing age.¹² Commonest endocrine disorder observed was hypothyroidism (23.8%). Mihci E et al, reported 11.88% of congenital hypothyroidism and further 37.62% becoming hypothyroid on follow up in their series.⁶ Four children (9.5%) were detected to have hypothyroidism in the newborn period and six diagnosed to have developed it during follow up. This emphasizes the need for a systematic checklist based follow up of these children and periodic parental education programs and support to the family. Diabetes and decreased fertility can also occur but not observed in the study.

Children with Down syndrome have an increased risk of developing leukemias, including acute lymphoblastic leukemia (ALL) and acute myeloid leukemia (AML).¹³ AML is as common as ALL in these individuals. Hasle H et al, estimate that the relative risk of acute leukemia in the first 5 years of life is 56 times that of individuals without Down syndrome.¹⁴ Two children developed ALL and received chemotherapy in this study.

More than 50% of the DS children in this series did not show malnutrition based on their weight for age. Only 9.5% had severe degree of malnutrition in the present study according to the IAP classification 1972. 61.3% had normal height for age according to Waterlow's classification. Severe stunting was noted only in 9.5%. Microcephaly was present in 65%.

Down syndrome being a genetic disease with longevity of life extending to more than forty years, this study has brought out only the issues related to children. Hence medical problems in a still large group of affected

population beyond the pediatric age group are not revealed by this study. A community based broader study may be needed to get a comprehensive picture and magnitude of the problem.

CONCLUSION

Down syndrome is a common genetic disorder with multisystem involvement. Congenital heart diseases, hypothyroidism and recurrent respiratory infections were the common medical problems identified in this study. Proper health education and provision for checklist-based screening for medical problems should be instituted for structured and comprehensive health care program for these special children. This certainly will improve their health status and as well as the quality of life.

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