Original Research Article

Parental awareness about medical comorbidities and their recommended evaluations in children with Down Syndrome: A pilot survey.

Abstract:

Down syndrome is associated with multiple comorbidities like growth delays, eye abnormalities, cardiovascular complications etc. They can be screened to detect early and can be intervened at an appropriate age. Professional organizations and other parents' support group can work towards streamlining the screening protocol and raising awareness.

Aim: To study the awareness about these comorbid conditions and their evaluations among parents of children with Down syndrome.

Method: A cross section questionnaire-based study pilot survey was conducted after taking due consent from the parents. It contained questions regarding whether the parents were aware of the increased risk in different organ systems and whether they knew the recommended timeline for screening.

Results: More than 90 percent of mothers were below the age of 35 years at the time of conception. More than half the parents knew about growth delays, eye, thyroid, cardiovascular complications. Less than 50% had awareness about DS specific growth charts, or the annual thyroid evaluations, while none knew the upper age limit to complete polysomnography evaluation.

Comment [paod1]: In the abstract we must find all the elements such as the objective of the research, the method used and the results obtained. You don't need to present it the way you did.No need to separate them into objectives and results. Do it all in one block of text. We can find them below in the different stages of your article

Key words: Parental awareness, knowledge, Down syndrome, comorbidities.

Introduction:

Down syndrome (DS) caused by trisomy of chromosome 21, is the most prevalent genetic disease worldwide and the most common genetic cause of intellectual disabilities, appearing in about 1 in 780 new-borns.[1]

While Down syndrome presents with a typical facial and morphological profile, it is also associated with growth issues and multiple medical comorbidities. In DS, reduced growth velocity and impaired growth spurts leads to short stature [2]. Children with DS have sensorineural hearing and mixed hearing loss. They are also prone to multiple attacks of acute otitis media and a majority develop chronic otitis media [3]. Obstructive sleep apnoea can lead to development of pulmonary hypertension or cor pulmonale [4]. DS has also been associated with different ophthalmological manifestations like strabismus, amblyopia, nystagmus etc [5]. Thyroid dysfunction is the most common endocrinal concern in DS [6]. Haematological abnormalities can range from minor cell line abnormalities like thrombocytopenia, polycythaemia to myelodysplastic syndrome and a high risk of leukaemia [7].

Most of the comorbid conditions can be detected early if screened regularly and can be treated either by medical or surgical intervention. Parenteral awareness about these conditions and the need for regular screening can decrease both the morbidity and financial burden on the families.

Aim of the study:

To conduct a pilot survey about the level of awareness amongst parents of DS children regarding the various common medical problems and the recommended frequency of evaluations.

Material and Methods:

The survey was a cross sectional and questionnaire-based study. It was administered on 23 parents of DS children who attended a comprehensive multidisciplinary medical camp conducted at Fernandez Child development centre, Hyderabad on World Down syndrome Day. The process was explained to the parents and consent was obtained. The questions were based on the Indian Academy of Pediatrics (IAP) guidelines to screen and evaluate the common medical problems in DS [8]. They were answered on an electronic tablet with a trained healthcare worker providing assistance and clarifications if required.The survey consisted of demographic factors of parents and categorical questions about whether the parents had knowledge about the different comorbidities and the timeline to screen them.

Results:

A total of 23 parents of children with DS were surveyed for the study. 14 (61%) mothers and 9 (39%) fathers answered the survey questions. 13(57%) of the children were male and 10 (43%) were female. Maternal age at the time of conception was equally distributed-in the 25-30 years and 31-35 years age groups with 10 mothers each (43%). Only 2 (9%) were above the age of 35 years and 1 (4.3%) was below the age of 25 years. Majority of the parents 13 (56.5%) had postgraduate education qualifications.

Out of 23 parents surveyed 21 (91%) were aware that children with DS can face delays in growth and development but only 6 (23%) knew the recommended frequency of checking height and weight annually and marking them in Down Syndrome specific growth charts.

Parents of 13 of the 23 children (56%) did not know that DS children are more prone to chronic ear infections and hearing loss. 14 parents (61%) knew the about the recommended annual ear evaluation.

More than half of the parents, 15 (68%) knew about the increased risk of eye problems like squint, cataract etc. 18 Parents (78%) knew about the recommended frequency of getting regular eye evaluations done i.e., annually till 5years, every 2 years till 12 years and every 3 years thereafter.

During the study,16 parents (70%) of the parents knew about the high incidence of thyroid conditions in DS. Only 9 (38%) were aware about the recommended annual testing of thyroid profile.

Around 70% of the parents knew about the high risk of a DS child being born or developing heart ailment, with 17 (74%) of them ready to get the ECG and echo done annually.

Although more than 80% of the parents knew about breathing/snoring issues leading to sleep disturbances, none of them were aware of the recommended need to get sleep studies (polysomnography) before the age of 4 years.

While less than 10 percent were able to name the various blood conditions that can affect children with DS, more than half (52%) were aware of the recommended annual complete blood count check to be done.

Most of the medical information that the parents received was either through medical personnel (doctor, nurses, or therapist) or a parent support group. A vast majority 96% of the parents wanted to get more medical information about DS. Almost 65% of them preferred to get the information in person as an advice from their treating doctor.

Discussion:

Gender ratio in DS is skewed to a male prevalence with a sex ratio 1.3:1 [9]. Our survey had a similar ratio of 13 males to 10 females. The chance of having a child with Down syndrome is present at any age but the risk increases exponentially after the age of 35 years. Still, most babies with Down syndrome are born to women under the age of 35 as this age group, inspite of having a lower risk, give birth more frequently than older women. In our survey 91% of the children were born to mothers who were less than 35 years and only 9% were above 35 years of age

Growth velocities of children with DS are markedly different compared to neurotypical healthy children **[**10]**.** Anthropometric measurements and plotting them on DS specific growth charts are recommended annually after the age of 1 year. In our survey, while more than 90 % of the parents knew that DS children might face growth delays, less than quarter of them knew about the recommended frequency of measurements or the availability of DS specific growth charts.

Acute and chronic otitis media is present in 20-40 percent of children with DS. Hearing loss is seen in almost three quarters of children with DS [3]. More than half of the parents did not know about the high proportion of ear problems, although around 60 percent of them knew the recommendation about the annual ear evaluation.

Children with DS have a high risk of developing ophthalmological complications like cataract, amblyopia etc [5]. A vast majority of the parents in our survey knew about this high risk and the recommended frequency of getting regular eye evaluations.

Thyroid dysfunction is the commonest endocrine abnormality in DS [6]. It can include congenital, subclinical, acquired hypothyroidism and hyperthyroidism. While almost three quarters of the parents know about high prevalence of thyroid problems, less than 40 percent knew the need to get yearly thyroid profile testing.

Congenital heart disease (CHD) is the leading cause of mortality and morbidity in DS with a prevalence of 40 to 60% [11]. More than 70% of the parents knew about the risk of CHD and the requirement for regular cardiovascular check-ups.

OSA is estimated to be prevalent in 30-80% of children with DS and universal screening through polysomnography is recommended by 4 years of age [12]. More than 80% of the parents surveyed knew about the complications of sleep apnoea but none of them knew that 4 years was the upper age limit to get polysomnography done.

DS children are prone to multiple haematological disorders ranging from cell line abnormalities like thrombocytopenia to Myelodysplastic syndrome [7]. Though more than half of the surveyed parents knew about the need to regularly check the blood counts less than 10 percent had awareness about the different disorders that could occur in them.

Its heartening to know that most of the parents wanted to know more about the different medical conditions in DS and preferred in person information from their doctors.

Conclusion: Many parents have a vague knowledge about the different medical conditions that might affect children with DS, but they do not have any specific knowledge about the recommended time period and type of evaluations to be done. It becomes imperative on the part of the paediatrician to be updated about the latest testing and evaluation guidelines and impart it to the parents during the regular visits. Anticipatory Guidance from the paediatrician plays an important role in raising awareness, screening. diagnosis and early intervention of the comorbid conditions in Down Syndrome

Limitations: The sample size was less as it was a pilot study and might not accurately reflect the results in a general population. A review whether the parents retained the knowledge of health comorbidities and timelines of screening could not be assessed.

Consent: Obtained before the start of the survey.

Ethical Approval: Since it was a pilot study, the name and other personal details of the subjects were not recorded. Hence ethical approval was not taken.

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