Optimizing Health Care for Individuals with Down Syndrome in Israel

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ABSTRACT: **Background:** Down syndrome is one of the most common chromosomal abnormalities. Children and adults with DS have significant medical problems and require life-long medical follow-up.

Objectives: To determine the adequacy of medical surveillance of individuals with DS as recommended by the American Academy of Pediatrics.

Methods: The study was conducted at a multidisciplinary center specializing in the care of DS during the period 2004–2006. At their first visit to the Center, caregivers of individuals with DS were questioned about the medical status of their child including previous evaluations. Medical records brought in by the parents were reviewed.

Results: The caregivers of 150 individuals with DS (age ranging from newborn to 48 years old, median age 5 years) were interviewed and the medical records were reviewed. The prevalence of specific medical problems differed between our population and the reported prevalence from other surveys. For example, 39.3% of our population had documented auditory deficits while the reported prevalence is 75%. For gastrointestinal and thyroid disease, the prevalence was higher in the studied population than that reported in the literature. In terms of compliance with the AAP recommendations, most children (94%) underwent echocardiography, but only 42.7% and 63.3% had been tested for auditory or visual acuity respectively. Only 36.3% over the age of 3 years had cervical spine films.

Discussion: Many individuals with DS are not receiving appropriate medical follow-up and the consequences of inadequate surveillance can be serious.

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Down syndrome is one of the most common genetic disorders in the world and the leading cause of mental retardation [1-3]. Although more than 80% of DS pregnancies can be diagnosed in prenatal tests, the rate of DS newborn deliveries has remained stable over the last decade – most likely due to advancing maternal age and cultural and religious objections to pregnancy termination [4,5]. The Israeli Ministry of Health estimates that the frequency of DS is 1:1000 live births [5]. Since there are approximately 150,000 live births annually in Israel, this translates to about 150 newborns with the syndrome.

DS is a chronic and complex medical condition that affects multiple systems including the central nervous system, and orthopedic, neurologic and endocrine systems. It has a high prevalence of congenital heart disease, dislocation of cervical vertebrae, and dysmorphic features. Children and adults with DS are at higher risk for developing disorders such as hypothyroidism, hearing and visual impairment, obesity, diabetes, dyslipidemia and presenile dementia. At the same time, individuals with DS face challenges related to education, employment and social integration [6-8]. Optimal care requires close follow-up and early intervention to minimize disease and dysfunction related to DS [3,9].

Optimal medical management of children with DS is one of the factors associated with improved level of functioning and quality of life [9]. To improve the care afforded to DS children and adults, various recommendations, including those formulated by the American Academy of Pediatrics, regarding diagnostic testing, follow-up, and the appropriate delivery of well-child care for DS have been published [9,10]. The degree of implementation of these recommendations varies from country to country, and the adult recommendations are less well developed than those for children [11].

In Israel, the management of DS is relatively decentralized with only a few centers providing multidisciplinary care [12]. Most individuals with DS are followed in community health management organizations, but the consistency of care and routine follow-up has not been evaluated in a systematic manner. It is possible that many of the recommended routine examinations for individuals with DS are not performed in a timely fashion.

The purpose of the present study was to determine whether and to what extent current recommendations regarding DS health care delivery are being implemented in Israel. We surveyed a convenience sample of subjects at their first visit to a DS clinic. The questionnaire contained items based on

DS = Down syndrome
AAP = American Academy of Pediatrics

the recommended protocol of diagnostic testing proposed by the American Academy of Pediatrics. The results of our survey indicate that a significant proportion of children had not undergone the diagnostic testing and evaluation that is currently recommended.

SUBJECTS AND METHODS

The study was conducted at the Hadassah Down Syndrome Medical Center during the period 2004–2006. The Center's primary goal is to provide comprehensive medical and social care to children and adults with DS. The Center provides comprehensive rehabilitative services, early diagnosis of potential disorders, and appropriate medical follow-up. It adheres to the recommendations formulated by the AAP with regard to surveillance and testing of DS children. Patients are scheduled for visits every 6–12 months (based on age) and are seen by a multidisciplinary staff consisting of pediatric subspecialists, rehabilitation experts, social workers and nurses. Patients also undergo a battery of tests to determine their level of function and mental status.

The patient population comprised patients with diagnosed DS and their parents or caregivers. After receiving an explanation, primary caregivers were queried about the medical condition and the care that their children received. Medical records and pertinent documents were also reviewed. The study received the approval of the Hadassah University Medical Center Institutional Review Board (Helsinki Committee).

THE QUESTIONNAIRE

The questionnaire comprised three sections. The first consisted of demographic questions pertaining to gender, age, ethnicity, age of parents, household members, and level of education of the subject. The second was a medical evaluation and included questions on current health status, prescribed medications, and ongoing medical, educational and social interventions. Prevalence rates of our population were then compared to prevalence rates of the same disorder reported in the medical literature [9,13]. The third section consisted of questions on the type and degree of medical surveillance offered to the patient. Specifically, families were asked about laboratory tests, radiographic studies, hearing and vision evaluations, and when their child underwent his/her last ophthalmologic and endocrine assessment. The last two sections were formulated to be consistent with the recommendations of the AAP (e.g., has your child been seen by an ophthalmologist in the past year?). Prior to the visit, caregivers were also asked to bring all medical documentation in their possession and to request summaries of laboratory tests performed at the patient's HMO.

HMO = health management organization

This information was reviewed by the study investigators and included in the surveillance data. Data from the survey were entered in an Excel spreadsheet (Microsoft).

RESULTS

During 2004–2006 we surveyed the parents or caregivers of 150 individuals with DS. The average age of the study population was 8.3 ± 8.9 years (range 0–48, median 5 years) and the mean maternal age was 33.6 ± 7.2 years. There was a predominance of males, with a male:female ratio of 1.5. In terms of ethnicity, approximately 10% of the families were of Arab ancestry.

Primary caregivers were questioned regarding specific medical problems affecting their child. Table 1 presents a breakdown of the reported diseases by system together with the reported prevalence as documented by the AAP. In some categories, there was an increased incidence in our population compared to other survey examples and this related to different inclusion criteria. For example, nearly all surveys and registries simply reported anatomic malformations in the category of gastrointestinal disease; in our survey we included both anatomic and functional disorders (e.g., recurrent abdominal pain, constipation, celiac). Similarly, under the category of thyroid dysfunction, we included children with overt hypothyroidism, subclinical hypothyroidism (elevated thyroid-stimulating hormone, normal thyroxin) and those who reported treatment with thyroid hormone in the past.

The number of disorders affecting the auditory and ophthalmologic systems was lower than the reported prevalence. This was most notable in the case of auditory defects, with a reported prevalence of 75% hearing loss (50–70% also have otitis media), whereas in our population only 39.3% of individuals had hearing loss and/or serous otitis media. Orthopedic problems also appeared to be underreported in our patient population, since the expected prevalence is 20% and only 8.7% of our subject population had orthopedic problems. With regard to cardiovascular disorders, the per-

Table 1. Prevalence of specific medical problems among Down syndrome individuals

System	No. of subjects affected (n=150)	Reported prevalence
Cardiovascular	90 (60.0)	50%
Auditory	59 (39.3)	75%
Ophthalmologic	69 (46.0)	60%
Orthopedic	13 (8.7)	20%
Gastrointestinal	69 (46.0)	13%
Thyroid	37 (24.7)	15%

Values are n (%)

Table 2. Medical follow-up of individuals with Down syndrom

Medical Investigations that were done in accordance with AAP recommendations	No.	%
Thyroid function tests	91	60.7
Hearing test	64	42.7
Eyes and vision check	95	63.3
Echocardiography	141	94.0
Cervical spine film (relevant to 91 subjects) › 3 years	33	36.3

cent of individuals in our clinic with cardiac anomalies was close to that reported in the literature (60% vs. 50%).

Primary caregivers were also asked about specific diagnostic tests that were performed on their children. Medical documents brought in by the parents were also reviewed. Table 2 demonstrates the number of individuals who underwent the prescribed surveillance as recommended by the AAP. For example, the AAP recommends that thyroid testing be done every 6 months until the age of 3 years and once a year thereafter. We found that only 60.7% of our total population had thyroid tests done in the past year. More distressing, many children had never had a thyroid evaluation after the newborn period. A significant proportion of the subjects had not been tested for thyroid according to the recommended time schedule. Despite the recommendation that hearing and visual testing be done every year, only 42.7% of our clinic population had a hearing test in the past year and 63.3% had a test for visual acuity. Similar to thyroid testing, a number of families reported that their children had never had formal audiologic, ophthalmologic or visual acuity examinations. Children with DS are at risk for cervical subluxation and the recommendations are to do a cervical spine radiological study at least once after the child has reached age 3. In our study, only a third of the children over the age of 3 years had cervical spine radiographs.

The highest level of compliance with the published recommendations occurred in early performance of echocardiograms. The AAP recommends that an echocardiogram be done soon after birth, and this was the case in 94.0% of our subjects.

DISCUSSION

Down syndrome is a prevalent genetic disorder with significant phenotypic variation and heterogeneity. Complications of DS can often be life threatening and are associated with significant mortality and morbidity. Not all the defects are present at birth and those that are, such as hearing and visual disturbances, may be difficult to detect at an early stage. From a different perspective, early intervention for sensory and endocrine disorders may be beneficial and improve outcome. Thus, timely and consistent follow-up is important for DS children, and scheduled routine follow-up is recommended by professional organizations [9,10].

The purpose of our study was to determine whether a selected population with DS in Israel, seen for the first time in a specialized clinic, underwent the appropriate surveillance recommended for children with DS in community-based clinics. Our results show that in specific disease subsets such as endocrinological and sensory organ surveillance, recommended surveillance was either not performed or not done in a timely manner. Potential factors related to inadequate surveillance are listed in Table 3. With regard to caregiver awareness, there are two possibilities. The first is that the recommended tests were done and the results were normal but the parents were not informed. The second is that the tests were not done at all, and the parents were unaware that their children needed to undergo specific evaluations. With regard to the first possibility, we made every effort to obtain all the available medical information by asking parents to talk to their child's physician and obtain the results of all testing done. Although it cannot be proved conclusively, the data from Table 1 would suggest that many of the surveillance tests were not performed, based on the fact that the prevalence of many of the commonly associated problems associated with DS were lower than the reported prevalence. While it is possible that the parents were informed that their children had sensory or endocrine problems but forgot about them (recall bias), it is our impression that parents when notified that their children had a hearing defect remembered this information and instituted appropriate treatment. This is also supported by data from follow-up screening, which showed that many patients did have specific diseases that had not been previously diagnosed. For example, all patients in our clinic were referred for audiologic exams, and the prevalence of hearing disorders among those who went for testing was similar to the prevalence among the general DS population (unpublished data).

There were some interesting discrepancies in our study. Nearly all caregivers recalled that their children had under-

Table 3. Surveillance of individuals with Down syndrome

Factors related to inadequate surveillance of individuals with DS
1. Lack of caregiver awareness regarding medical needs of DS individuals
2. Testing done but parents unaware
3. Medical professionals not cognizant of special requirements of DS
4. Health care systems not structured to provide appropriate DS care
5. Non-allocation of resources for DS surveillance programs
6. Negative caregiver attitudes towards DS
7. Negative attitude of health care professionals regarding DS

Potential interventions
1. Educational programs targeting care givers and health care providers
2. Adoption of a DS surveillance and follow-up program by pediatric associations and dissemination to health care providers
3. Provision of a health card to individuals with DS listing testing schedule and results
4. Greater utilization of multidisciplinary centers providing DS care

gone echocardiography, and the prevalence of congenital heart disease was even higher than the reported prevalence among DS individuals. In contrast, the frequency of auditory testing was fairly low (42.7%) and the reported incidence of hearing deficits was lower than the known prevalence. One possibility is that the group surveyed for this study was non-representative of the general DS population. Individuals attending our clinic were either self-referred or sent by physicians and may have had more serious manifestations of DS. Another possible explanation for this discrepancy is that not all health care providers are aware or agree with the AAP's recommendations for DS. For example, the AAP recommends cervical spine films at age 3 to rule out the possibility of cervical spine subluxation. However, the England and Ireland Down's Syndrome Medical Interest Group advises against such a policy because of the unreliability of such radiographs and the low frequency of cervical spine instability [10].

A third explanation for this discrepancy is the nature and timing of specific modes of health care delivery in Israel. Children born with DS often undergo cardiac echocardiography while still in the nursery as a matter of routine, and there is a high degree of awareness among the nursery staff regarding cardiac problems associated with DS. Hearing and visual testing, on the other hand, is often done as part of the government Mother and Child Surveillance Program (well-baby clinics), but there are no formal or comprehensive guidelines on the management of children with DS. Most other interactions with the health care system in Israel focus on the ill child or adult, and diseases such as hypothyroidism are not routinely tested. Also, few multidisciplinary centers that provide comprehensive care for children and adults with DS have systematic surveillance programs.

What are the implications of a lack of systematic monitoring for children and adults with DS? The most dangerous, immediate and life-threatening disorders are related to cardiovascular or anatomic gastrointestinal problems, and as our data show, surveillance is adequate for these disorders. Other disorders such as hearing loss or endocrinopathies may be diagnosed after a significant delay. Although the delay may not always result in irreversible damage, early diagnosis and intervention is very important. For example, recognition of a hearing loss detected during the period of speech development is important since early intervention may prevent speech pathology, which is often a problem in DS children [14]. Likewise, hypothyroidism in the first year of life can result in intellectual disability, and this too is preventable with early detection based on routine surveillance. Often it is difficult to diagnose thyroid disorders in individuals with DS because the clinical manifestations of the syndrome may mask the clinical manifestations of thyroid dysfunction [15]. It is also important to remember that DS children face significant physical and intellectual challenges, and it is important to detect even disorders that are not exclusive to the DS population as these diseases may have a disproportionate affect on the DS child. Moreover, because of the delays in speech development and comprehension, recognizing common manifestations such as visual acuity defects may also be delayed. For example, we detected a severe refractory error in a 1.5 year old child and cataracts in other children about which the parents were unaware. Finally, many disorders such as congenital heart disease can have a disproportionate impact on physical and cognitive development in DS.

Attitudes towards DS are changing and there is greater recognition that many DS children can lead normal and productive lives provided that there are early medical, rehabilitative and educational interventions. Recent advances and progress in understanding the molecular basis of DS holds out the promise that there will be better therapeutic interventions in the future [16]. Individuals with DS face many challenges, and with appropriate medical surveillance some of the difficulties encountered by DS children and adults can be mitigated [17,18].

In summary, children and adults with DS in Israel are not always followed appropriately. Several potential remedies for this problem are listed in Table 3. One solution is to heighten the awareness of both the medical community and parents of DS children regarding appropriate medical surveillance. Similar to the cards that parents receive from the governmental Mother and Child Surveillance Programs, caregivers of DS patients could be given a card with a list of tests that must be done as part of a DS surveillance program. Another potential solution is to refer DS children and adults to a multidisciplinary center that provides comprehensive medical follow-up.

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