The perception of otolaryngology-related diseases among parents of children with Down syndrome in Jeddah, Saudi Arabia

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ABSTRACT

Objectives: To assess the perception of otolaryngology-related diseases among parents of children with Down syndrome (DS).

Methods: A cross-sectional survey design. A questionnaire was distributed to parents of children with DS. The parents were attending an event aiming to raise awareness that was organized on World Down Syndrome Day on the 21st of March 2013, organized by Princess Al-Jawhara Center of Excellence in Research of Hereditary Disorders, King Abdulaziz University, Jeddah, Saudi Arabia. The complete dates of study were March to May 2013.

Results: Questionnaires were completed and perceptions regarding 10 common otolaryngology-related diseases were obtained from 78 parents. The data shows that most of the parents were not familiar with these otolaryngology-related diseases.

Conclusion: Many parents were not aware of some of the otolaryngology related conditions affecting children with DS, and did not have regular follow-up. Perception was highly affected by the presence of the condition in the children. Awareness of otolaryngology-related diseases affecting children with DS should be raised.

Down syndrome (DS), trisomy 21, is a genetic disorder caused by the existence of all or part of a third copy of chromosome 21.[1] It is the most common chromosomal abnormality in humans.[2] The Center for Disease Control approximates that around one in every 691 babies born in the United States each year is born with DS.[3] The incidence in Saudi Arabia is one in every 554 births (1.8 per 1000).[4] It is typically associated with a delay in cognitive ability and physical growth and a particular set of craniofacial characteristics that leads to otolaryngologic (ORL) problems. This includes cerumen impaction, chronic middle ear infections and effusion due to eustachian tube abnormalities, hearing loss, obstructive sleep apnea, as well as problems with chronic rhinitis and sinusitis.[5] However, there is a lack of prior studies in the literature assessing the perception and awareness of these problems among parents of children with DS. For this reason, on World Down Syndrome Day, Princess Al-Jawhara Center of Excellence in Research of Hereditary Disorders organized a public awareness campaign for DS, titled "My Rights in My Society". This study was conducted during the campaign to evaluate the perception and knowledge of ORL problems among parents of children with DS; to increase their awareness and highlight the importance of regular follow-up to detect and prevent these conditions and their complications. We also aimed to raise awareness among physicians of the need to implement regular follow-up programs and proper treatment for children with DS.

Methods. This study adopted a survey cross-sectional design. The questionnaires were completed using a convenience-sampling approach. The participants included in the study were parents of children with DS who attended a public awareness campaign for DS. The campaign was organized on World DS Day, on the 21st of March 2013, by Princess Al-Jawhara Center of Excellence in Research of Hereditary Disorders, King Abdulaziz University, Jeddah, Saudi Arabia. Other visitors that attended the event who were not parents of children with DS were excluded. Approval was obtained from the Research Ethics Committee in King Abdulaziz University Hospital. The researchers developed the survey, and then experts in the field of Otolaryngology, Head and Neck surgery reviewed it. The questionnaire was not validated. The survey included demographic variables and questions that measured the perception of ORL-related diseases among parents of children with DS. No specific demographic questions regarding the participants were included since investigating the characteristics of the sample was not the main aim of the study. To insure confidentiality of the collected information, no personal data were obtained from the participants. Consent was taken verbally from parents who visited the ORL awareness booth. They were asked to take part in this study by completing the questionnaire. We asked participants to choose one or more symptoms or conditions they believed that children with DS might have or develop from

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an ORL prospective. The study was conducted in the Otolaryngology, Head and Neck Surgery Department of King Abdulaziz University, Faculty of Medicine, Jeddah, Kingdom of Saudi Arabia.

The data were entered in Microsoft Excel, and then imported to the Statistical Package for Social Sciences (IBM SPSS Statistics for Windows, Version 21.0. Armonk, NY: IBM Corp.) to be analyzed. Descriptive analysis was produced using the SPSS program. The frequency and percentages were reported for the categorical variables. Our study used the multiple response frequency analysis to interpret the common choices among the sample. The percentage (%) of responses indicates what percentage of the total number of symptoms mentioned is contained in each category or symptom. Cross-tabulation for multiple responses was used to compare based on the frequency and percentages, the perception among those with a child experiencing a condition and a child not experiencing that condition.

Results. In total 78 questionnaires were completed. All parents that attended the Otolaryngology booth agreed to participate in the study. The data displayed in **Table 1** illustrates the percentage of children who had previously visited an ORL physician (78.2%), how many of them had regular follow-up (70.5%), and the follow-up intervals. It also shows the percentage of children who were currently experiencing ORL related symptoms or conditions (83.3%), how many of them underwent ORL operations (30.8%), if any of them had complications after those surgeries (12.5%), and an outline of these complications. The symptoms that were perceived (symptoms that parents identified as conditions that might affect children with DS) and experienced (symptoms that the parents reported present in their children) included ear wax, otitis media, hearing loss, difficulty understanding words, delayed speech, difficulty in learning, difficulty in interacting with others, snoring, obstructive sleep apnea, and allergic rhinosinusitis. The frequency as shown in **Table 2** indicates how many respondents mentioned each symptom. Note that the total of responses adds up to more than the sample size (78) because subjects ticked more than one symptom. A comparison was made to illustrate the relationship between the frequencies on perception and the presence of the disease only among those who perceived it, excluding parents who reported that their child experienced the condition but did not know that it was a common condition affecting many children with DS in general as shown in **Table 3**.

Table 1 - Demographic questions, number of responses (n), and percentages (%) among parents of children with DS.

Demographic questions	n	(%)
Visited an ORL physician before		
Yes	61	(78.2)
Follow-up (n=61)		
Yes	43	(70.5)
Period of follow-up (n=43)		
Only when needed	17	(39.5)
Every month	6	(14.0)
Every 2 months	1	(2.3)
Every 3 months	4	(9.5)
Every 4 months	1	(2.3)
Every 6 months	9	(20.9)
Once a year	6	(14.0)
Child experiencing ORL symptoms or conditions		
Yes	65	(83.3)
ORL operations		
Yes	24	(30.8)
Any complication after operation (n=24)		
Yes	3	(12.5)
Type of complications (n=3)		
Velopharyngeal insufficiency	1	(33.3)
Weight loss	1	(33.3)
Sleep disturbance	1	(33.3)

DS - Down syndrome, ORL - otolaryngology

Table 2 - Frequency (%) of perceived and experienced conditions by children with DS.

Symptoms	Perceived		Experienced	
	n (%)			
Ear wax	35	(14.6)	21	(14.4)
Otitis media	18	(7.5)	10	(6.8)
Hearing loss	23	(9.6)	18	(12.3)
Difficulty understanding words	15	(6.3)	9	(6.2)
Delayed speech	38	(15.9)	32	(21.9)
Difficulty in learning	22	(9.2)	12	(8.2)
Difficulty in interacting with others	18	(7.5)	9	(6.2)
Snoring	29	(12.1)	16	(11)
Obstructive sleep apnea	17	(7.1)	11	(7.5)
Allergic rhinosinusitis	24	(10.0)	8	(5.5)

DS - Down syndrome, OSA - Obstructive sleep apnea

Table 3 - Frequency of experienced conditions among children with DS who perceived it.

Conditions	Does your child have this condition?		Total
	Yes	No	
Ear wax	31	4	**35**
Otitis media	18	0	**18**
Hearing loss	21	2	**23**
Difficulty understanding words	14	1	**15**
Delayed speech	34	4	**38**
Difficulty in learning	19	3	**22**
Difficulty in interacting with others	17	1	**18**
Snoring	27	2	**29**
Obstructive sleep apnea	17	0	**17**
Allergic rhinosinusitis	21	3	**24**
Total	**64**	7	**71**

DS - Down syndrome, OSA - Obstructive sleep apnea

Discussion. Down syndrome is the most common chromosomal abnormality in humans, it is well known that ORL problems are more common in children with DS and the effect of these problems is significant on those children. After evaluating the parents' questionnaires it is suggested that many of them are not too familiar with some of the ORL-related conditions that can occur in their children as shown in **Table 1**, although we used lay terms whenever possible and tried to stay away from medical terms. Some parents may not have been given details by their Otolaryngologist of the ORL problems that their children might develop or are currently going through. As noted in **Table 1**, 21.8% of the 78 parents we interviewed mentioned that they had not visited an Otolaryngologist before, and out of the 61 children who had visited an Otolaryngologist 29.5% did not have regular follow-up in the clinic, which might have had a significant influence on their knowledge regarding these common conditions that their children might develop or are currently suffering from without medical attention.

We compared our results with data provided by Princess Al-Jawhara Center of Excellence in Research of Hereditary Disorders. Out of 111 individuals with DS, in which the vast majority were children (males: 63, females: 48) (age range: 1-36 years) (mean age: 6.5 years), registered in the center we found that 30% did not have regular follow-up visits with the ORL department. When compared with our results at a significance level of 0.05, there was no significant difference regarding follow-up between the 2 groups (z=1.11106, *p*-value=0.267). We also noted that the follow-up visits for 17 (39.5%) of those children who had visited an Otolaryngologist were only when they were symptomatic, which indicates that many of their parents were not aware of the need for regular follow-up visits. The follow-up intervals displayed in **Table 1** were variable among those 43 who were seen in different institutions.

In our study, we found that the most commonly perceived condition by the parents enrolled in the study was delayed speech (15.9%), followed by ear wax (14.6%), snoring (12.1%), rhinosinusitis (10%), hearing loss (9.6%), difficulty in learning (9.2%), otitis media (7.5%), difficulty interacting with others (7.5%), obstructive sleep apnea (7.1%), and difficulty in understanding words (6.3%). Regarding the most commonly perceived condition, namely, speech delay (15.9%), we also found it to be the most commonly experienced condition. Speech delay is caused by multiple factors in children with DS. One factor is the degree of cognitive deficits, with 80% of the individuals showing moderate retardation, which leads to language deficits, particularly in expressive language and syntax, as well as poor speech intelligibility.[6] Another factor found to be linked to speech delay is hearing loss.[6] Reported hearing loss was found to be as high as 78%,[7] yet only 41% of the parents reported that their children suffered from it in our study. This high rate may also be attributed to inadequate treatment of chronic otitis media with effusion that was perceived by 23% of the parents, and yet only 12% claimed that their children had it in our study. Roizen et al[8] showed that there was an increased incidence and a high rate of under-diagnosis and under-treatment of otitis media in children with DS.

Regarding earwax, the second most commonly perceived condition in our study (14.6%), only 26% of the parents reported that their children had this condition. Several studies attributed the higher incidence of cerumen impaction among DS children to the fact that 40-50% of the newborns with DS have stenotic ear canals,[9] and these narrowed canals predispose the children to cerumen impaction. Snoring, the third most commonly perceived issue (12.1%) is a symptom of airway obstruction and obstructive sleep apnea. Obstructive sleep apnea has a high incidence among DS children, estimated at 31-63%[10,11] (20% in our study). It could be a result of the small airways obstructed with glossoptosis, enlarged tonsils and adenoids, hypotonia, or obesity.[5]

The marked difference between the incidence of ORL issues in our study and other studies could be attributed to the use of parental reporting to determine the experienced conditions, as these parents who attended the event represent a cross-sectional sample of the local community with varied educational backgrounds. This clearly underestimates the presence of these conditions among the children and emphasizes the need for proper counseling of parents by primary and secondary care practitioners on the complications that their children might develop due to the lack of proper follow-up, which in itself is a key factor contributing to a higher rate of under-diagnosis.

The parents' perception was compared with the presence of these conditions among their children. We found out that perception of a condition is highly affected by the presence of this condition in the children. Among 38 parents who believed that delayed speech is a common condition affecting children with DS, only 4 of the children's parents did not mention delayed speech as a condition affecting their children.

As for ear wax that was perceived by 35 parents, only 4 of them mentioned that their children did not have this condition. In other conditions like obstructive sleep apnea and otitis media only the parents who mentioned that those conditions affected their children perceived it. A comparison between the other conditions is displayed in **Table 3** as well.

A limitation that faced our study was the lack of prior research assessing the perception of ORL problems among parents of children with DS. Other limitations included the lack of justification for the sample size prior to the event, not including the parents' demographic data that might have helped in identifying the most misinformed parents in the population, and the use of parental reporting to establish the presence of ORL problems in the children as this may affect the results due to memory bias.

In conclusion, many of the parents were not aware of some of the otolaryngology related conditions affecting children with DS and did not have regular follow-up. Moreover, many of the parents who had visits to the ORL clinic only did so when their children were symptomatic. Perception was highly affected by the presence of the condition in the children. More awareness of the common conditions affecting children with DS from an otolaryngology perspective is needed. In addition, physicians need to educate the parents on the conditions affecting, or those that might affect, their children and emphasize the need for regular follow-up visits. We highly recommend the use of the American Academy of Paediatrics' guidelines for health supervision of children with DS.[12] In future studies, including the parents' demographic data might help identify the most misinformed segment of the population and facilitate better awareness among those parents.

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