Bronchoscopic Findings in Down Syndrome Children with Respiratory Problems

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Background: *Down syndrome (DS) can affect the upper and lower respiratory tract in a number of ways and disorders of other systems can impact upon respiratory function, giving rise to a wide variety of respiratory manifestations.*
Objective: *To investigate the frequency, associated conditions, and type of airway anomalies in DS children with respiratory problems.*
Material and Method: *Twenty-nine children with DS were evaluated for airway anomalies, with indications of atelectasis, stridor, and recurrent or persistent pneumonia, using flexible fiberoptic bronchoscopy (FB). The children were assessed retrospectively.*
Results: *Results showed high frequency of associated conditions, which was found in 19 children. The most common associated condition was congenital heart disease. Other associated conditions were hypothyroid and duodenal atresia. Endoscopic findings showed several airways anomalies, including laryngomalacia, tracheal bronchus, and subglottic stenosis. Three patients had multiple airway anomalies and three had normal endoscopic findings.*
Conclusion: *DS children with respiratory problems have high frequency of airway anomalies and FB, which is a useful diagnostic procedure. A thorough understanding of the airway anomalies will aid in the evaluation and management of DS children with respiratory problems.*

Keywords: *Down syndrome, Bronchoscopy, Airway anomalies*

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Down syndrome (DS), trisomy 21, is the most common chromosomal disorder, with an incidence of approximately 1 in 800 live births. Children with DS are more likely to have medical problems, including congenital heart defects, Hirschprung disease, duodenal atresia, and thyroid dysfunction[1]. DS can affect the upper and lower respiratory tracts in a number of ways, giving rise to a wide variety of respiratory manifestations including airway obstruction, recurrent respiratory infections and wheeze[2].

There is currently insufficient data available regarding airway anomalies in children with DS. Early identification of the cause of respiratory problems is important for appropriate management. In this study, we assessed the frequency, associated conditions, and the types of airway anomalies and their clinical presentations. Flexible fiberoptic bronchoscopy (FB), which provides accurate assessment of pediatric airway disorders, was used to investigate airway anomalies.

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Material and Method

Patients

The present study assessed 29 DS children who underwent airway evaluation using FB at Queen Sirikit National Institute of Child Health (QSNICH) between November 2009 and November 2013. Their hospital records were reviewed and the following data were collected: demographic characteristics, associated conditions, indication for FB assessment and FB findings.

FB assessment

The patients underwent FB under sedation with topical anesthesia in the operation room. The Olympus FB with outer diameter of 2.8 mm and 3.5 mm was used. Hemodynamic and respiratory statuses were continuously monitored and supplementary oxygen was given during the procedure. The airway was examined for intrinsic abnormalities, intraluminal lesions, evidence of extrinsic compression and abnormal airway

dynamics.

Results

Review of the demographic data showed that the study population was made up of 18 boys (62%) and 11 girls (38%), with the median age being 11.9 months (2 months-4 years). Primary indications for the procedure were atelectasis in 4 patients, stridor in 10 patients, and recurrent or persistent pneumonia in 15 patients (Table 1).

The most common associated conditions were congenital heart disease, which was found in 13 patients. Other associated conditions were hypothyroid and duodenal atresia. Ten patients had no associated conditions (Table 2).

FB data showed a high frequency for airway anomalies in children with DS (89.6%). Airway anomalies included laryngomalacia in 5 patients, tracheal bronchus in 4 patients, subglottic stenosis in 3 patients, laryngotracheomalacia in 3 patients, tracheal stenosis in 2 patients, external compression of trachea in 2 patients, pharyngomalacia in 1 patient, external compression right upper lobe bronchus in 1 patient, tracheomalacia in 1 patient, abnormal branching right upper lobe bronchus in 1 patient. In three patients, multiple airway anomalies were found, laryngomalacia and tracheal bronchus in 1 patient, subglottic stenosis and tracheomalacia in 1 patient and tracheal stenosis, laryngomalacia and tracheal bronchus in 1 patient. Three patients had a normal FB examination (Table 3).

Discussion

Respiratory problems are an important cause of morbidity and mortality in children with DS. In a review of death certificates, respiratory illnesses (aspiration, pneumonia and influenza) were the second most common cause of death for children with DS aged up to 19 years, and the most common cause across all age groups[3]. In another study involving patients with DS under the age of 3 years, respiratory illnesses were the most common primary reason for admission[4].

FB assessment is safe and provides a means of assessing airway anomaly, which contribute to a variety of respiratory problems. More recently, the use of the pediatric FB has increased the rate of diagnosis airway anomalies in children with DS. The authors found high frequency of airway anomalies in children with DS as compared to one study about FB findings in non-DS children who had atelectasis, stridor and recurrent pneumonia (89.6% vs. 35%). Children with DS may have abnormal or obstructed airways for a variety of reasons (Table 4)[2].

Both laryngomalacia and tracheomalacia are associated with DS and may present with stridor. Laryngomalacia has been found to be the most common cause of upper airway obstruction in children with DS under 2 years[5]. Laryngomalacia may occur in association with other anomalies of the airway[6].

In the present study, the authors found 2 out of 7 children with DS (28.5%) diagnosed laryngomalacia also had other airway anomalies e.g. tracheal bronchus,

Table 1. Primary indications for FB (n = 29)

Respiratory problems	n	%
Atelectasis	4	13.8
Stridor	10	34.5
Recurrent or persistent pneumonia	15	51.7

Table 2. Associated conditions in children with DS and respiratory problems (n = 19)

Associated conditions	n	%
Congenital heart disease	13	68.5
Congenital heart disease with hypothyroid	3	15.9
Congenital heart disease with duodenal atresia	1	5.2
Hypothyroid	1	5.2
Duodenal atresia with hypothyroid	1	5.2

Table 3. Types of airway anomalies (n = 26)

Airway anomalies	n	%
Laryngomalacia	5	19.3
Tracheal bronchus	4	15.5
Subglottic stenosis	3	11.6
Laryngotracheomalacia	3	11.6
Tracheal stenosis	2	7.7
External compression of trachea	2	7.7
Pharyngomalacia	1	3.8
Tracheomalacia	1	3.8
Abnormal branching right upper lobe bronchus	1	3.8
External compression right upper lobe bronchus	1	3.8
Subglottic stenosis and tracheomalacia	1	3.8
Laryngomalacia and tracheal bronchus	1	3.8
Tracheal stenosis, laryngomalacia and tracheal bronchus	1	3.8

Table 4. Airway problems in children with DS

Upper airway problems related to phenotypic features	Macroglossia
	Midface hypoplasia
	Narrow nasopharynx
Associated conditions affecting upper airway	Adenotonsillar hypertrophy
	Choanal stenosis
Structural problems of larynx/trachea	Laryngomalacia
	Narrow trachea
	Tracheomalacia
	Subglottic stenosis
Other contributing factors	Obesity
	Hypotonia

tracheal stenosis. Tracheomalacia results from a malformation of the cartilage ring that only partially surrounds the circumference of trachea or from localized softening as a result of the chronic pulsatile effect of a vascular ring or abnormal big vessel[6]. However, endoscopic appearance of our patients was consistent with the former mechanism.

Tracheal bronchus is a congenital anomaly seen in DS consisting of an aberrant or accessory bronchus arising from the trachea. While this may be an incidental finding, it may also be associated with respiratory disease, particularly recurrent right upper lobe pneumonia. Mclaughlin et al[7] reported a 2% incidence in a series of over 400 pediatric bronchoscopies, which reflect the results of others in an otherwise normal population. In our study, 15.5% DS patients were diagnosed tracheal bronchus that was higher when compared to non-DS patient. Significant morbidity associated with tracheal bronchus in children with DS could be related to poor handling of secretions and/or aspiration. The authors may consider tracheal bronchus a significant airway anomaly in DS due to the high incidence of associated respiratory morbidity, especially in infants, where the recumbent position is the rule during most of the day.

In addition to the smaller trachea seen in patients with DS[8], there may be increased incidence of subglottic stenosis. Miller et al[9] felt that the relatively high proportion of patients with DS among those undergoing laryngotracheal reconstruction when compared with the overall incidence of DS suggested an increased incidence of subglottic stenosis in DS patients. This has important implications for clinicians who might intubate children with DS. It is recommended that a smaller endotracheal tube be used than would be expected for the patient's weight[8]. Congenital tracheal stenosis has been reported in patients with DS[10,11]. In patients with DS and congenital tracheal stenosis, the most common type appears to be segmental "hour glass" stenosis[12]. However, most cases of subglottic stenosis appear to be acquired post intubation; in one series of 17 patients with DS and subglottic stenosis, the stenosis was felt to be congenital in only four cases[9]. It is not clear whether the suggested increase in incidence of subglottic stenosis in children with DS is due to an intrinsic increased risk of subglottic stenosis, or because children with DS are more likely to undergo surgery (e.g. for heart or gastrointestinal defects) and, therefore, intubation[9]. However, in another series of findings at bronchoscopy in patients with respiratory symptoms, the incidence of tracheal stenosis was not significantly increased in patients with DS compared with those without[6].

The authors found 7.7% of children with DS diagnosed external trachea compression. Tracheal narrowing due to external compression is also associated with congenital heart disease that is related to dilated vessels, anomalous vessels or cardiac enlargement[13]. The authors also found that lobar or segmental bronchial variants are recognized in children with DS.

Conclusion

In summary, DS children with respiratory problems have a high incidence of airway anomalies. FB provides a means of assessing these airway anomalies. Respiratory problems are an important cause of morbidity and mortality in children with DS. Awareness of airway anomalies in children with DS and respiratory problems may improve disease/ treatment outcomes.

Potential conflicts of interest

None.

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ผลการตรวจส่องกล้องระบบทางเดินหายใจในเด็กกลุ่มอาการดาวน์ที่มีปัญหาระบบทางเดินหายใจ

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ภูมิหลัง: กลุ่มอาการดาวน์สามารถส่งผลกระทบต่อระบบทางเดินหายใจส่วนบนและส่วนล่างได้หลายอย่าง และยังมีความผิดปกติอื่นร่วมด้วยที่สามารถส่งผลกระทบต่อการทำงานของระบบการหายใจ ทำให้เกิดอาการแสดงของระบบหายใจได้หลายอย่าง

วัตถุประสงค์: เพื่อศึกษาถึงความถี่ ความผิดปกติที่พบร่วมด้วยและชนิดของความผิดปกติของทางเดินหายใจในเด็กกลุ่มอาการดาวน์ที่มีปัญหาระบบทางเดินหายใจ

วัสดุและวิธีการ: เด็กกลุ่มอาการดาวน์ที่ได้รับการส่องกล้องตรวจทางเดินหายใจเนื่องจากปอดแฟบ, หายใจเสียงดัง stridor และปอดอักเสบที่เป็นซ้ำหรือปอดอักเสบที่ไม่ดีขึ้นเมื่อให้การรักษาที่เหมาะสม โดยการศึกษารวบรวมข้อมูลย้อนหลัง

ผลการศึกษา: ในระยะเวลาของการศึกษาเด็กกลุ่มอาการดาวน์ 29 ราย ได้รับการตรวจหาความผิดปกติของทางเดินหายใจเนื่องจากปอดแฟบ, หายใจเสียงดัง stridor และปอดอักเสบเป็นซ้ำหรือปอดอักเสบที่ไม่ดีขึ้น เมื่อให้การรักษาที่เหมาะสม เด็กกลุ่มอาการดาวน์ 19 ราย พบความผิดปกติร่วมด้วยโรคหัวใจพิการแต่กำเนิด เป็นความผิดปกติที่พบร่วมด้วยได้บ่อยที่สุด นอกจากนี้ยังพบความผิดปกติที่พบร่วมด้วยอื่น ๆ เช่น ภาวะพร่องไทรอยด์ฮอร์โมนและลำไส้ส่วนดูโอดีนัมตีบ ความผิดปกติของทางเดินหายใจที่พบได้แก่ กล่องเสียงอ่อนตัว (5/26), tracheal bronchus (4/26), ทางเดินหายใจบริเวณใต้กล่องเสียงตีบแคบ (3/26), กล่องเสียงและหลอดลมใหญ่อ่อนตัว (3/26), หลอดลมใหญ่ตีบแคบ (2/26), หลอดลมใหญ่ถูกกดจากภายนอก (2/26), คอหอยอ่อนตัว (1/26), หลอดลมใหญ่อ่อนตัว (1/26), การแตกแขนงของหลอดลมปอดกลีบขวาบนผิดปกติ (1/26), หลอดลมปอดกลีบขวาบนถูกกดจากภายนอก (1/26), ทางเดินหายใจบริเวณใต้กล่องเสียงตีบแคบและหลอดลมใหญ่อ่อนตัว (1/26), กล่องเสียงอ่อนตัวและ tracheal bronchus (1/26), หลอดลมใหญ่ตีบแคบ, กล่องเสียงอ่อนตัวและ tracheal bronchus (1/26) เด็กกลุ่มอาการดาวน์ 3 ราย พบความผิดปกติของทางเดินหายใจมากกว่า 1 อย่าง และมีเด็กกลุ่มอาการดาวน์ 10 ราย ตรวจไม่พบความผิดปกติของทางเดินหายใจ

สรุป: เด็กกลุ่มอาการดาวน์ที่มีปัญหาระบบทางเดินหายใจพบความผิดปกติของทางเดินหายใจได้สูง การส่องกล้องตรวจทางเดินหายใจมีประโยชน์ช่วยในการวินิจฉัย การมีความเข้าใจเกี่ยวกับความผิดปกติของทางเดินหายใจช่วยในการประเมินและให้การรักษาเด็กกลุ่มอาการดาวน์ที่มีปัญหาระบบทางเดินหายใจ