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Tracheostomy Dependance Patterns in Children with 22q11 Deletion Syndrome

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Abstract

Introduction: Due to the medical complexity of 22q11.2 deletion syndrome, some people require tracheostomy. To date, there is little in the literature regarding tracheostomy for these people. It was our aim to better delineate patients with 22q11DS who require tracheostomy and assess outcomes, including decannulation and complications.

Methods: This is a retrospective chart review of patients in our 22q Center's repository. Inclusion criteria were a diagnosis of 22q11.2DS with a current or previous history of tracheostomy.

Results: 170 charts were reviewed and 10 children (5.9%) met inclusion criteria. All children had CHD and 3 had history of cleft palate. Mean age at tracheostomy was 6.22 months (range 2 months – 15 months). The most common indication for tracheostomy was cardiac/respiratory failure (n=7). Nine children were decannulated with a mean tracheostomy duration of 3.94 years (range 1 year – 8 years); 1 child passed away at 5 years of age prior to decannulation. Decannulation required laryngotracheal reconstruction in 3 children.

Conclusion: All children requiring tracheotomy had a history of CHD. Most children were successfully decannulated, although it often took many years and additional procedures to facilitate decannulation. This information may aid preoperative counseling for families of children with 22q11.2 deletion syndrome requiring tracheostomy.

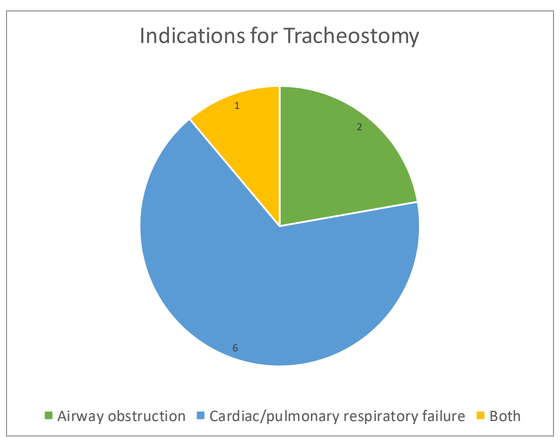
Introduction

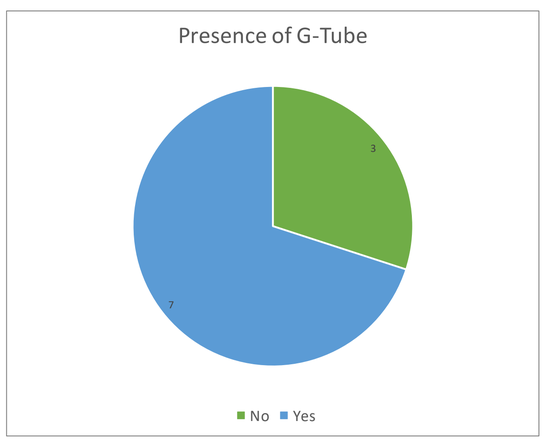
22q11.2 deletion syndrome (22q11.2DS) is the most common chromosomal microdeletion syndrome occurring in approximately 1 in 2148 live births with an autosomal dominant pattern of inheritance (1). Affected individuals present with a wide range of symptoms and conditions including congenital heart disease (CHD), cleft palate, velopharyngeal insufficiency, immunodeficiency, developmental delay, and hypocalcemia.

Airway findings in 22q11.2DS have been well described in the literature. Due to the medical complexity of this syndrome, some people require tracheostomy. To date, there is little in the literature regarding tracheostomy dependance in this population. It was our aim to better delineate people with 22q11.2DS who require tracheostomy and assess outcomes, including decannulation and complications.

Methods and Materials

After obtaining Institutional Review Board approval, a retrospective chart review of all children in our 22q Center's repository was conducted. Inclusion criteria were a diagnosis of 22q11.2DS with a current or previous history of tracheostomy. Demographic data was collected which includes current age, gender and age at tracheostomy. Past medical history was collected including history of CHD, cleft palate, indication for tracheostomy, complications, successful decannulation, duration of tracheostomy, length of follow-up, history of laryngotracheal reconstruction, G-tube dependance, and mortality.





Results

A total of 170 charts were reviewed and 10 children (5.9%) with 22q11.2DS underwent tracheostomy. All children had CHD and 3 had history of cleft palate. Mean age at tracheostomy was 6.22 months (range 2 months – 15 months). Nine children were decannulated with a mean tracheostomy duration of 3.94 years (range 1 year – 8 years). Decannulation required laryngotracheal reconstruction (LTR) in 30% of children (n=3).

Flexible laryngoscopy while inpatient revealed bilateral vocal cord paralysis (VCP) in 2 children and unilateral VCP in 2 children.

History of G-tube placement was present in 80% (n=8) of children. Of these, 3 children had their G-tube removed with an average G-tube duration of 5.5 years (range 4 years – 7 years). Four children still had G-tube at the time of last follow up.

One child passed away at 5 years of age prior to decannulation and G-tube removal.

Table 1. Clinical features and outcomes for 22q11.2DS children requiring tracheostomy

Subjects	1	2	3	4	5	6	7	8	9	10
Gender	F	M	F	M	M	M	F	F	F	F
Age at tracheostomy (months)	7	Unk^	9	2	6	3	2	15	8	4
Indication for tracheostomy*	O	Unk^	O	B	C	C	C	C	C	C
Duration of Tracheostomy	7 yr	Unk^	8 yr	13 mo	Unk^	4 yr	1.5 yr	4 yr	10 mo	5 yr
Successful decannulation	Y	Y	Y	Y	N	Y	Y	Y	Y	Y
Need for LTR	Y	Y	N	N	N	N	N	N	N	Y
Need for G-tube	N	N	Y	Y	Y	Y	Y	Y	N	Y
Presence of cardiac defect	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y
Presence of cleft palate	N	Y	N	N	N	N	Y	N	Y	N
Deceased	N	N	N	N	Y (5 yr)	N	N	N	N	N

*O: Airway obstruction, C: Cardiac/pulmonary respiratory failure, B: Both
^Unk: Unknown

Discussion

Previous studies have described airway abnormalities in children with 22q11.2DS. Sacca et al. noted that 71% of people with 22q11.2DS who underwent microlaryngoscopy and/or bronchoscopy were found to have an airway abnormality (2). While not all airway anomalies result in a need for tracheostomy, in our cohort, 5.9% of children required tracheostomy. It's certainly notable that 100% of the patients with tracheostomy also had CHD. Sacca et al. also found a high correlation between CHD and tracheostomy. Future studies are needed to further investigate this subset of patients with 22q11.2DS, CHD, and significant airway disease (2).

It is encouraging that in this study 90% of children were eventually decannulated. Decannulation required multiple surgical airway interventions, including LTR, in 30% of children, highlighting the need for tracheostomy patients to be closely followed by an otolaryngologist.

Feeding difficulties are common in this population of children with 22q11.2DS and tracheostomy; 80% of children in our cohort required a G-tube at some point demonstrating the need for feeding therapy to be closely involved in these children's care.

Conclusions

The present study describes patterns of tracheostomy in children with 22q11.2DS. Interestingly, all children requiring tracheotomy had a history of CHD. Most children were successfully decannulated, although it often took many years and additional procedures to facilitate decannulation. This information may aid preoperative counseling for families of children with 22q11.2 deletion syndrome requiring tracheostomy.

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References

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