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Otolaryngologic surgery in children with trisomy 18 and 13

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ABSTRACT

Objectives: Trisomy 18 and 13 are the most common autosomal trisomy disorders after Down syndrome. Given the high mortality rate (5–10% one-year survival), trisomy 18 and 13 were historically characterized as uniformly lethal and palliation was the predominant management approach. Management strategy has shifted with recognition that through medical and surgical intervention, children with trisomy 18 and 13 can achieve developmental milestones, live meaningful lives, and exhibit long-term survival. Otolaryngologic surgery in children with trisomy 18 and 13 has not been described. The objective of this article is to describe the role of the otolaryngologist in the management of children with trisomy 18 and 13.

Methods and materials: Retrospective cohort analysis of the surgery registry for the Support Organization for Trisomy 18, 13 and Related Disorders for otolaryngologic surgeries reported from 1988 through June 1, 2014.

Results: In the database of approximately 1349 children, 1380 procedures were reported, 231 (17%) of which were otolaryngologic. The most common otolaryngologic procedures were tympanostomy tube placement (57/231, 25%), cleft lip repair (40/231, 17%), tracheostomy (38/231, 16.5%), tonsillectomy and/or adenoidectomy (37/231, 16%), and cleft palate repair (30/231, 13%). Of the ten most common procedures reported, four were otolaryngologic.

Conclusions: Seventeen percent of procedures performed in children with trisomy 18 and 13 were otolaryngologic, highlighting the significant role of the otolaryngologist in the treatment of these patients. Surgical intervention may be considered as part of a balanced approach to patient care.

1. Introduction

Trisomy 18 and trisomy 13, also known as Edwards syndrome and Patau syndrome, respectively, are the most common autosomal trisomy disorders after Down syndrome (trisomy 21) [1]. The overall prevalence of trisomy 18 and 13 in the United States, including stillbirth, elective pregnancy termination, and live birth is estimated to be 1/2500 and 1/6400, respectively [2]. Diagnosis is most often made in the prenatal period based on screening for advanced maternal age, maternal serum markers, amniocentesis, and sonographic abnormalities [3]. The vast majority of cases are full or partial trisomy with fewer than 5% of patients demonstrating mosaicism.

The major clinical features of trisomy 18 include facial dysmorphism with micrognathia, ear abnormalities, cardiac defects, rocker bottom feet, and overlapping digits. Major clinical features of trisomy 13 include holoprosencephaly, midline facial defects, cleft lip and palate, cardiac defects, and polydactyly [4]. In both syndromes, the majority of children exhibit significant developmental and psychomotor delay.

There is a very high rate of fetal loss following prenatal diagnosis of trisomy 18 and 13. An estimated 72% of trisomy 18 fetuses diagnosed at 12 weeks do not survive to term. The analogous statistic for trisomy 13 is 49% [5]. Median postnatal survival for trisomy 18 receiving palliative care alone is 3 to 15 days with 5 to 10% of children surviving beyond one year [3]. Survival figures in trisomy 13 are very similar [1]. The foremost causes of death are central apnea, cardiac failure, and respiratory insufficiency [3,6].

The high mortality rate in children with trisomy 18 and 13 historically characterized these syndromes as uniformly lethal and the conventional therapeutic approach was predominantly palliative. However, children with trisomy 18 and 13 may achieve social

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and physical developmental milestones including social smiling, recognizing and interacting with family members, laughter, signing, sitting upright, self-feeding, and unassisted walking [7–10]. There have been case reports of long-term survivors of full trisomy living up to 19 years old [11]. Furthermore, surviving children live meaningful and happy lives as determined by their families [10,12].

While historic mortality rates have been very high, recent studies have demonstrated that provision of intensive care and surgical intervention significantly improves survival, up to 27% at one year [13,14]. In light of these data and the growing understanding of the meaningful lives led by these patients, the approach to treatment has changed, care providers are offering life-extending interventions, and families are electing to proceed. The interventions are performed by a variety of pediatric subspecialists including otolaryngologists.

Few publications address surgical intervention in children with trisomy 18 and 13 and only one study in the English literature describes otolaryngologic treatment [6,10]. The primary objective of this study was to describe the role of the otolaryngologist in the management of children with trisomy 18 and 13 by examining the nature and prevalence of procedures performed in this population.

2. Materials and methods

The Support Organization for Trisomy 18, 13 and Related Disorders (SOFT) is a support group for families of patients with trisomy 18, 13 and other related chromosomal disorders. SOFT maintains a surgery registry with information on surgical procedures reported by parents of children with trisomy 18 and 13 from the late 1980s to present. This is an open-access online database (accessible at http://trisomy.org/wp-content/uploads/2014/05/SurgTypesReport5-13-2014.pdf) and patient information has been de-identified. In a retrospective cohort analysis of the SOFT surgery registry, information was collected on all otolaryngologic surgeries performed from 1988 through June 1, 2014. Data collected included otolaryngologic procedures performed among patients with complete/partial trisomy and mosaicism. Procedures were included as "otolaryngologic procedures" if taught in United States Otolaryngology residency training and regularly performed by Otolaryngologists. It is understood by the authors that some of these procedures, especially cleft lip/palate repair and tracheostomy, may have been performed by physicians practicing in other specialties.

3. Results

The database includes approximately 4400 children. 1380 procedures were reported on children with trisomy 18 or 13. The most common procedures overall were gastrostomy (246/1380, 17.8%), fundoplication (60/1380, 6%), and tympanostomy tube insertion (57/1380, 4.1%).

Of the 1380 procedures, 231 (16.7%) were otolaryngologic. The most common otolaryngologic procedures performed in descending order were tympanostomy tubes (57/231, 25%), cleft lip repair (40/231, 17%), tracheostomy (38/231, 16.5%), tonsillectomy and/or adenoidectomy (37/231, 16%), and cleft palate repair 30/231, 13%). Other otolaryngologic surgeries accounted for 29/231 including ear canaloplasty, choanal atresia repair, cholesteatoma removal, mastoidectomy, salivary gland removal, and supraglottoplasty.

Trisomy 13 patients had higher rates of cleft lip and palate repair than patients with trisomy 18 (5.0% vs. 1.6% and 4.2% vs. 0.9%, respectively). There was no difference in rate of tympanostomy tube placement (5.4% vs. 3.4%), tonsillectomy and/or adenoidectomy (2.1% vs. 3.0%), or tracheostomy placement (2.7% vs. 2.8%).

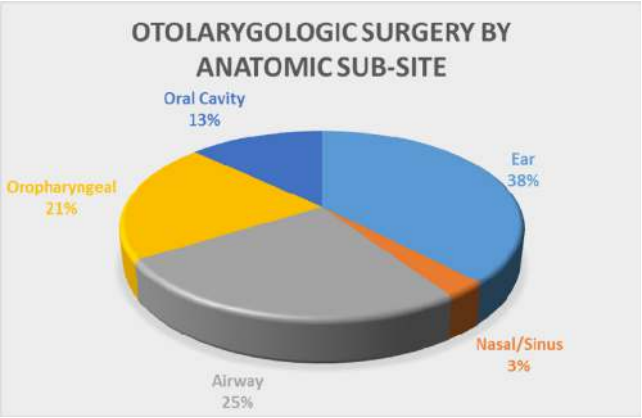

Fig. 1. Otolaryngologic surgery in trisomy 18 and 13 by anatomic sub-site.

The proportion of otolaryngologic procedures performed when categorized by anatomic location was as follows: 38% ear surgery, 25% airway, 21% oropharyngeal, 13% oral cavity, 3% nasal/sinus, and 0.4% salivary glands (Fig. 1).

When looking at procedures performed among all specialties, 4 of the 10 most common procedures performed in patients with trisomy 18 and 13 were otolaryngologic procedures: tympanostomy tube insertion (4.1%), cleft lip repair (2.9%), tracheostomy (2.8%), and tonsillectomy and/or adenoidectomy (2.7%) (Table 1).

4. Discussion

Trisomy 18 and 13 are two of the most common trisomy syndromes [1]. Although both of these disorders have a high mortality rate, they are not uniformly lethal as previously characterized. As care providers begin to understand that life-sustaining treatment may be warranted in certain cases, more trisomy 18 and 13 patients are becoming surgical candidates, particularly because the provision of intensive care and surgical intervention has recently been shown to significantly improve survival rates [13,14]. Otolaryngologists are being called upon to offer initial life-sustaining treatment as well as procedures to improve quality of life. However, up to now, the role of the otolaryngologist in the treatment of these patients has not been well defined.

Table 1
Most common procedures performed in patients with trisomy 18 and 13 out of 1380 procedures. VSD = ventricular septal defect, PDA = patent ductus arteriosus, ASD = atrial septal defect, T&(A) = tonsillectomy and (adenoidectomy).

Procedure	Number	Percentage
Cardiac		
VSD repair	55	4.0
PDA ligation	44	3.2
ASD repair	36	2.6
Gastrointestinal		
Gastrostomy	246	17.8
Fundoplication	80	5.8
Orthopedic		
Spinal fusion	33	2.4
Otolaryngologic		
Ear tubes	57	4.1
Tracheostomy	38	2.8
Cleft lip	40	2.9
T&(A)	37	2.7

Data previously reported in the literature describing otolaryngologic surgery performed on children with trisomy 18 and 13 only reviewed cleft palate repair and tracheostomy. Nelson et al. [6] reported that tracheostomy comprised 3.8% of overall procedures performed on trisomy 18 and 13 patients and cleft palate repair comprised 2% of overall procedures performed. In our study, we find a similar prevalence of these two procedures; tracheostomy comprised 2.8% of all procedures and cleft palate repair comprised 2.2% of all procedures.

In our study, approximately 17% of reported surgeries in children with trisomy 18 and 13 were otolaryngologic. The most common otolaryngologic procedure was pressure-equalizing tubes, accounting for 4.1% of all surgeries. In the general pediatric population, pressure-equalizing tubes account for 20% of ambulatory surgeries [15]. Tonsillectomy comprised 2.7% of surgeries in children with trisomy 18 and 13 while adenotonsillectomy comprises 16% of ambulatory surgeries in a general pediatric population [16]. The lower rates of ear tubes and adenotonsillectomy in the study population may reflect the high degree of medical complexity leading to greater selectivity of practitioners when evaluating surgical candidacy.

When compared to trisomy 18, trisomy 13 patients had higher rates of cleft lip and palate repair (5% vs. 1.6% and 4.2% vs. 1%, respectively) and tympanostomy tube placement (5.4% vs. 3.4). These findings are consistent with the increased prevalence of cleft palate in children with trisomy 13. Higher rates of tympanostomy tube placement among trisomy 13 patients may be attributed to eustachian tube dysfunction among cleft palate patients. In our study, tracheostomy comprised nearly 3% of surgeries in children with trisomy 18 and 13 indicative of the shifting trends in management of these patients as more providers offer life-sustaining treatments.

Several recent studies revealed that surviving children with trisomy 18 and 13 are happy, enrich the lives of their families, and provide meaningful quality time with caregivers [10,12]. As care providers, our understanding of trisomy 18 and trisomy 13 has been defined by the morbid pictures of neonates depicted in medical textbooks. These are in stark contrast to the photographs of Trisomy 18 and 13 children that can be found on the SOFT website that depict these children enjoying meaningful lives with their families. As our perspectives are being redefined, so is our approach to the management of these patients. Our current study reveals that the otolaryngologist is already playing a major role in the surgical care of these patients.

The rarity of these diseases makes prospective data collection challenging, however, acquiring data from a patient reported database has significant limitations. The primary limitation is a reporting bias; all procedures in the database are reported by parents of patients that may result in omitted or erroneously entered information. Furthermore, a depletion of the susceptible bias likely results in greater exclusion of phenotypically severe children as these children have higher mortality rates and are therefore not added to the registry [17]. This leads to a selection bias for patients with milder phenotypes that live longer and are more likely to have otolaryngologic surgeries performed. We are therefore likely overestimating the proportion of otolaryngologic surgeries among all children with trisomy 18 and 13.

5. Conclusions

The otolaryngologist has a significant role in the care of patients with trisomy 18 and 13. Otolaryngologists are encouraged to contribute to ongoing dialogue with regard to the management of children with trisomy 18 and 13 and participate in multidisciplinary care teams. Surgical intervention should be considered as part of a balanced approach to patient care.

Conflict of interest statement

None.

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