CONCLUSIONS: 22q11.2 (Velocardiofacial and DiGeorge) Deletion Syndrome is relatively common (1 in 2,000 children). Typical facial features may not be obvious so otolaryngologists may be first to diagnose the syndrome. Worse than usual Eustachian tube dysfunction (ETD), velopalatine insufficiency (VPI), global delays, and history of cardiopulmonary outflow tract anomalies should alert the surgeon to consider genetics evaluation/FISH testing. Common clinical features include the need for multiple ear surgeries despite ear tubes, speech delay/history of feeding problems suggestive of VPI, and persistent laryngotracheomalacia. Immune problems and infantile hypocalcemia are also common due to thymic hypoplasia.

OBJECTIVES: To describe presentation/outcomes in 22q11.2 patients requiring ENT surgery.


RESULTS: 114 of 571 patients referred to otolaryngology underwent 240 operations.

Ear surgery:
- 57/114 (50%) required tubes (BMT)
- 35/57 (61%) had multiple BMT
- 19/57 (33%) residual perforations
- 57 (9%) retained tubes
- 1 acquired ear canal atresia

Palate-related surgery:
- 36/57 (63%) with ear problems had velopalatine insufficiency (but only 6 had overt cleft palate)
- 12 of these patients underwent tonsillectomy +/- adenoïdectomy for breathing problems and/or to prepare nasopharynx for posterior pharyngeal flap
- Partial adenoïdectomy done in most to preserve speech quality or to avoid injury to medialized nasopharyngeal carotid artery (2/12 patients)

Airway surgery:
- An overlapping cohort of 104 children required microlaryngoscopy/bronchoscopy (MLB) because of stridor, VPI with feeding/speech/voice issues, sleep apnea, and/or airway compression due to conotruncal cardiovascular defects
- 74/104 (71%) had airway anomalies
- 36% trachomalacia
- 16% subglottic stenosis
- 26% laryngomalacia
- 21% glottic web
- 21% tracheobronchomalacia
- Tracheotomy required in 22/67 (30%) who had cardiac and airway anomalies

Children with 22q11.2 deletion are of all races and have variable facial features. More common are small or squared upper ears, hypertelorism, hooded eyes, asymmetric smile, stenotic ear canals, broad nasal dorsum, and bulbous nasal tip.

Many children have VPI and low muscle tone leading to hypernasal speech and persistent ETD requiring multiple ear procedures.

Common airway problems include anterior glottic webs and tracheomalacia which often are exacerbated by underlying cardiac problems.

Patients with 22q11.2 syndrome are missing a segment of genes on chromosome 22. Although 22q11.2 is almost as common as Down syndrome, patients present with variable clinical features, so it is often under diagnosed. It is inherited in an autosomal dominant fashion, but most cases occur randomly. Many patients have multiple otolaryngologic problems requiring interventions. Awareness of this syndrome and early therapies improve patient outcomes, so pediatric specialists have developed an international study group, “22q11.2 Society”, to provide resources for clinicians and as a forum for further research.