Dental Caries and Malocclusion in Patients with 22q11.2 Deletion and Duplication Syndromes

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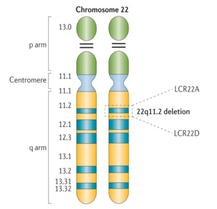
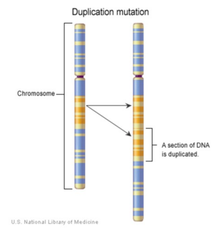
Background

- **22q11.2 Deletion Syndrome (22q11.2DS)** is a congenital disorder that occurs in **1 in 2,148 live births** and **1 in 992 pregnancies**
  - Most common microdeletion syndrome
  - Most common cause of syndromic cleft palate
  - Second most common cause of heart defects and developmental delays
  - Included Phenotypes:
    - DiGeorge Syndrome
    - Velocardiofacial Syndrome
    - Conotruncal Anomaly Face Syndrome
    - AD Opitz G/BBB Syndrome
    - Sedlackova Syndrome
    - Cayler Cardiofacial Syndrome
  - Reported oral characteristics include:
    - Cleft Palate
    - Dental Caries
    - Enamel Hypoplasia
    - Malocclusion





- There is little known about the oral characteristics regarding **22q11.2 duplication syndrome (22q11.2DupS)**
  - The prevalence of 22q11.2DupS has been reported in **1 in 850 pregnancies**.
  - Associated with a range of phenotypes including congenital heart defects, vision abnormalities, growth failure

- **Question Posed: What are the differences in orofacial and dental anomalies associated with 22q11.2DS and 22q11.2DupS?**

Methods

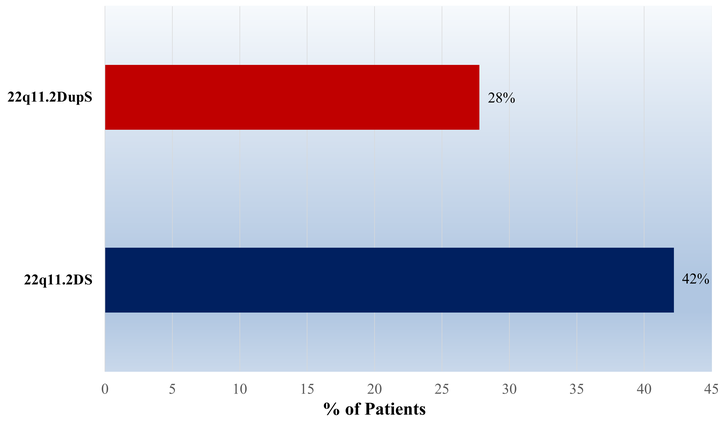
- **Retrospective Chart Analysis** of patients diagnosed with 22q11.2 DS or 22q11.2DupS and seen at the Children's Hospital of Philadelphia in the following clinics:
  - "22q and You" Clinic
  - Department of Speech and Language Pathology
  - Craniofacial and Special Needs Orthodontics Clinic

- **Recorded Findings:**
  - Age
  - Gender
  - Genetic Anomaly (22q11.2DS or 22q11.2DupS)
  - History of Congenitally Missing Teeth
  - History of Cleft Lip/Cleft Palate/Both
  - Other diagnosed syndromes
  - Occlusion (Class 1, Class 2, Class 3)
  - History of Crowding
  - History of Anxiety
  - History of Caries
  - History of Snoring

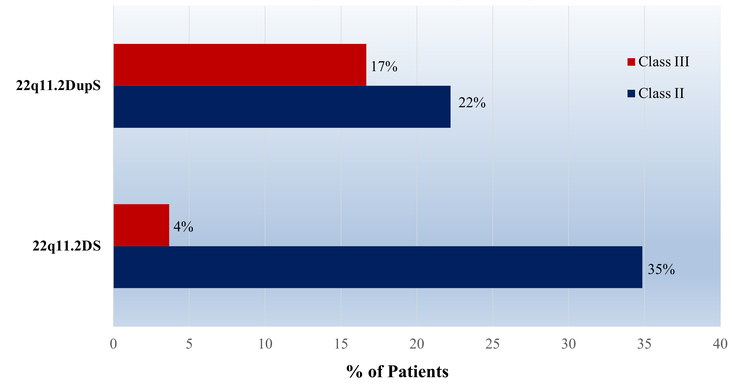
- Once patient charts were identified, clinical notes from the clinical genetics team, speech language pathologists, plastic surgery, and craniofacial orthodontics clinic were reviewed.
- In the instances where information regarding a specific topic was unavailable, we noted that the information was unreported.

Results

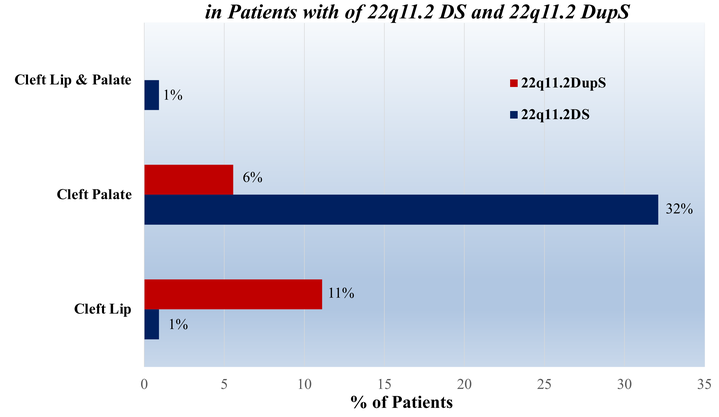
Caries Experience in Patients with 22q11.2 DS and 22q11.2 DupS



Class II or Class III Malocclusion in Patients with 22q11.2 DS and 22q11.2 DupS



Cleft Lip, Cleft Palate, or Cleft Lip & Palate in Patients with of 22q11.2 DS and 22q11.2 DupS



Results (cont.)

Distribution of Recorded Findings in Separated 22q11.DS and 22q11.2DupS cohorts

Recorded Findings	22q11.2DS (n=109)	22q11.2DupS (n=18)
Gender Distribution	59 F 50 M	8 F 10 M
Average Age	9 years old	8 years old
Caries Experienced	42%	28%
Overbite (Class II Malocclusion)	35%	22%
Underbite (Class III Malocclusion)	4%	17%
Crowding	32%	6%
Cleft Palate	32%	6%
Cleft Lip	1%	11%
Cleft Lip and Cleft Palate	1%	0%
Diagnosis of other syndromes	7%	22%
Missing Teeth	5%	6%
Snoring	42%	33%
Anxiety	32%	28%

Conclusions

- This is the largest sample size of any of the studies in reported literature; however, further chart review is needed to increase overall sample size. Data analysis can also be strengthened by increasing the sample size of the duplication cohort.
- Due to the sample size discrepancy between the two cohorts, statistical comparison cannot be completed.
- Due to incomplete evaluation/history of dentition by non-dental personnel, we predict the incidence of caries and malocclusion is lower in this sample size than it would be if dental evaluation/history was completed by a dentist.
- From the data collected, it is shown that caries prevalence is high in the 22q11.2DS and 22q11.2DupS population. Proper involvement of dentistry in a multidisciplinary care group is vital to these patients' overall well-being.

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