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## Background

### 22Q 11.2 Deletion Syndrome:

- Most common microdeletion syndrome (now estimated at 1 in 2,148 live births and 1 in 992 pregnancies)
- 2nd most common cause of heart defects and developmental delays
- **Included Phenotypes:** *DiGeorge Syndrome, Velocardiofacial Syndrome, Conotruncal Anomaly Face Syndrome, and many others*

### Enamel hypoplasia (EH) :

- Quantitative developmental defect of enamel resulting from disruption of ameloblast activity during the secretory phase of amelogenesis.
- Characterized by a reduced amount of enamel
- Affects 6-38 % of general population
- Enamel hypoplasia and hypomineralization prevalent in 22q11.2DS
- Enamel defects increase caries risk and treatment burden
- Potential link between systemic disease and enamel development

## Study Aims

To evaluate the prevalence of enamel defects in patients with 22q11.2DS and identify associated systemic findings

## Methods

**Retrospective chart review** conducted at the Children's Hospital of Philadelphia via the 22Q and You clinic and Craniofacial and Special Needs Orthodontics clinic of 132 patients diagnosed with 22q11.2 DS.

- Without Enamel Hypoplasia: n = 92
- With Enamel Hypoplasia n= 40

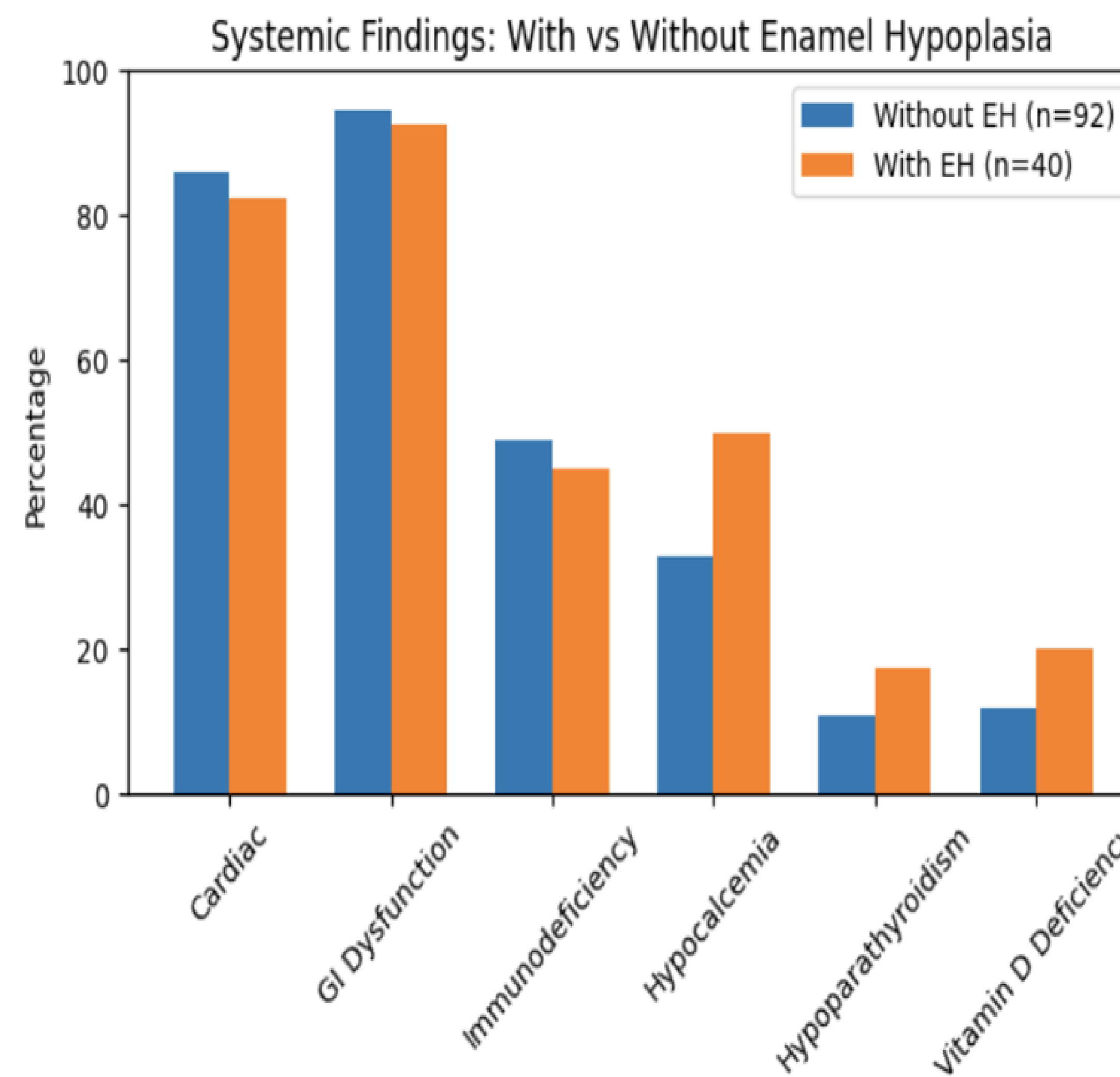
### Systemic findings:

- Cardiac anomalies
- Immune dysfunction
- History of hypocalcemia
- Hypoparathyroidism
- GI conditions
- Vitamin D deficiency

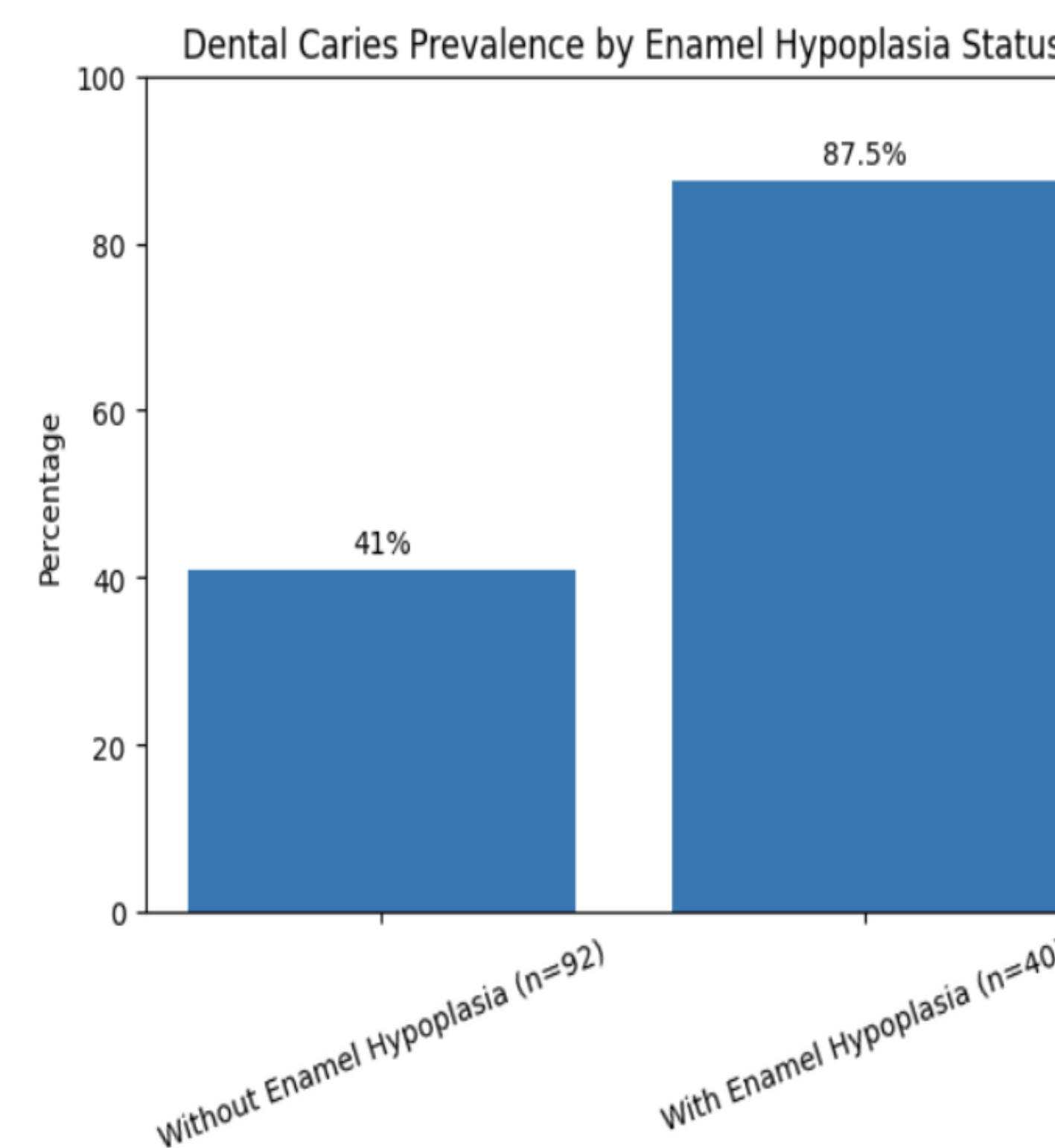
### Dental Findings:

- Diagnosis of Enamel hypoplasia
- Diagnosis of Dental caries
- Oral Hygiene

## Results

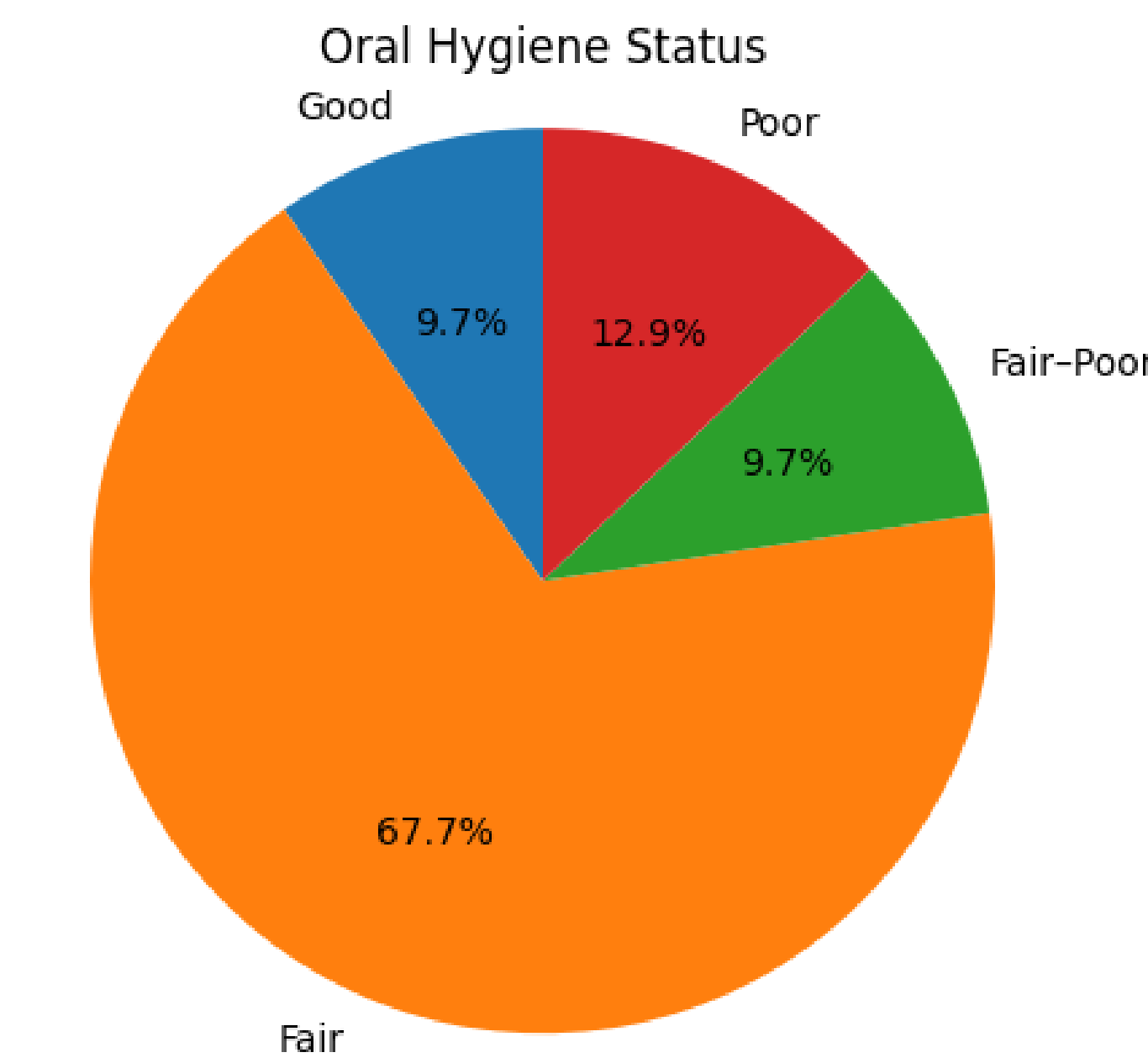


- 40 out of 132 patient presented with EH.
- Both cohorts demonstrate significant systemic involvement, particularly cardiac and gastrointestinal findings.
- Immunodeficiency rates were comparable between groups.



- Among patients without enamel hypoplasia, **41%** had documented caries.
- In contrast, **87.5%** of patients with enamel hypoplasia had caries.

## Results (continued)



- Most patients with Enamel Hypoplasia demonstrated fair oral hygiene.

## Discussion

- 30% of patients with 22Q deletion had enamel hypoplasia
- Hypocalcemia, hypoparathyroidism, and vitamin D deficiency were more prevalent in patients who had EH compared to patients without EH
- Despite the high caries prevalence, **most patients demonstrated fair rather than poor oral hygiene**, indicating that factors beyond hygiene alone may contribute to caries risk.

## Conclusion

- Enamel hypoplasia appears to be an **under-recognized dental phenotype in 22q11.2 deletion syndrome**.
- Enamel hypoplasia clusters with higher systemic disease burden
- Systemic comorbidities (hypocalcemia, hypoparathyroidism, and vitamin D) may contribute to **altered enamel development and increased caries susceptibility**, independent of oral hygiene status
- Patients with multiple systemic conditions may benefit from earlier dental surveillance and enhanced preventive strategies

## References

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