ORAL MANIFESTATION OF TRISOMY 21- A REPORT AND REVIEW

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ABSTRACT
Downs syndrome or trisomy 21 is a genetic disorder characterised by abnormal physical and mental growth. Individuals affected with such anomalies usually present a typical facial profile, with systemic manifestations. Herein we report a case with review on oral manifestation and changes in a patient with trisomy 21.

KEYWORDS: Downs syndrome, Trisomy, Chromosome, Oral manifestation.

INTRODUCTION
Trisomy 21 is a genetically inherited condition characterised by delayed physical growth, characteristic facial profile, and mild to moderate intellectual disability. [1] Usually parents of the affected individuals are genetically normal. [2] The Manifestations of downs syndrome was first described in the year 1866 by John Langdon Down. [3] The craniofacial and oral features involved in Down syndrome include brachycephaly, small nose associated depressed nasal bridge, narrow or small maxilla, high arched palate and tongue with fissures and papillary hypertrophy. [4]

CASE REPORT
A 25 year old patient came to the department of oral medicine and radiology with a chief complaint of deposits in the teeth. Medical history was non-contributory. Extraoral examination revealed characteristic facial profile with intercanthal distance of 35mm (Figure 1A). Intraoral examination revealed deep grooves on the dorsal aspect of the tongue which were painless, Suggestive of Fissured tongue. (Figure 1B) Gingiva was soft with deposits on the teeth, localised bleeding on probing was evident. Maxillary anterior teeth were missing. (Figure 1C) Teeth morphology was altered suggestive of Microdontia (Figure 1D) Correlating the intraoral and extraoral findings a Provisional diagnosis of Downs Syndrome/ Trisomy 21 was given. Patient was referred to the respective departments for the needful.

Table 1 – Extraoral and intraoral findings in a downs syndrome patient[1,2,3,4,5,6]

<table>
<thead>
<tr>
<th>Extraoral findings</th>
<th>Intraoral findings</th>
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<tbody>
<tr>
<td>Microcephaly</td>
<td><strong>Hard tissue:</strong> Dental caries, Microdontia, Anodontia, Hypodontia, High arched palate, Narrow palatal arch.</td>
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<tr>
<td>Flattened face</td>
<td><strong>Soft tissues:</strong> Aphthous ulcers, Bald tongue, Fissured tongue, Gingivitis, Periodontitis, Oral candidiasis, Halitosis.</td>
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<tr>
<td>Small nose</td>
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<td>Depressed nasal Bridge</td>
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<tr>
<td>Wide neck</td>
<td></td>
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<td>Almond shaped eyes</td>
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<tr>
<td>Dysplastic small ears</td>
<td></td>
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<td>Hypertelorism</td>
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FIGURE LEGEND.

Figure A: Measurement of intercanthal distance
Figure B: Fissured tongue
Figure C: Missing Maxillary Anterior Teeth
Figure D: Change in tooth morphology

DISCUSSION

Downs syndrome being a genetic alteration a diagnosis of which can be given by only correlating the appearance and manifestations related to it.\(^3\) It’s also called as trisomy 21 because 95% of the affected individual have extra chromosome.\(^5\) Delay in starting to speak, which only occurs at around four years of age, on average, slowing the pace of learning to read and write.

Trisomy 21 exhibits characteristic features which helps in identifying the syndrome (Table: 1) Differential diagnosis considered for trisomy 21 are congenital hypothyroidism, Mosaic trisomy 21 syndrome, partial trisomy 21, Robertsonian trisomy 21, Zellweger syndrome or other peroxisomal disorders.\(^7\)

CONCLUSION

Assessment, monitoring and parental counselling plays a major role in maintaining the oral health of patients with trisomy 21. Dental clinician and every medical professional should be able to identify this genetic disorder, so that the best care could be provided during the patient’s dental visit.

REFERENCES


