REVIEW ARTICLE

Association of Down Syndrome (DS) with Morphology and Anomalies of Teeth: A Concise Narrative Review

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Abstract

Trisomy 21 causes Down syndrome (DS), a genetic condition marked by unique dental and craniofacial abnormalities. These abnormalities significantly impact oral function, appearance, and overall quality of life. An example of this tendency is seen in cases where DS children erupt their first molar at 8 years and 9 months as opposed to the average rise in age between 6 and 8 years. The most prevalent kind of consequence for individuals with DS who have unfavourable diseases of the middle part of their faces is facial deformities. This review investigates the relationship between DS and dental anomalies, with a focus on tooth morphology, number, eruption patterns, and structural defects. It seeks to summarise existing studies, point out knowledge gaps, and emphasise therapeutic implications for oral healthcare. Databases like PubMed and Google Scholar were used to assess peer-reviewed literature. Included were studies on dental abnormalities in DS, such as hypodontia, microdontia, delayed eruption, enamel hypoplasia, and malocclusions, that were published between 2000 and 2025. A characteristic of DS is dental abnormalities, which call for specialist oral therapy to address both functional and cosmetic issues. Improving results requires multidisciplinary care, individualised treatment programs, and early diagnosis and action. General colonisation, dental health, and other medical issues are all positively impacted by dental surgery. In contrast to simpler cognitive subjects, IQ might occasionally demonstrate a reduced ability to handle more complicated cognitive activities. To investigate genetic causes and create focused management plans, more research is required.

Keywords: *Dental anomalies, hypoventilation, chromosomes, permanent dentition, hypodontia, and craniofacial dysplasia.*

INTRODUCTION

Historical Background of Down Syndrome

John Langdon Down (English doctor) was the person who coined the term mongolism in 1866 to describe Down syndrome (DS); however, Jerome Lejeune (French doctor) discovered that the extra number of Chromosome 21 was the cause of it. He went on to name the syndrome as chromosome 21 disease because of this. Chromosomal abnormalities, the most significant of which are the group which includes DS, which occurs as a result of an extra chromosome copy for chromosome 21, are among them. This is the most frequent type of condition, and it is commonly referred to as trisomy 21 or DS. The intrinsic hallmark of DS is the extra chromosome 21, which is the only chromosome that is present in all human cells [1-6].

Tooth Eruption in Down Syndrome

The child with DS has a gradual transition to their permanent dentition as opposed to an abrupt conversion, which is common for children without mental disabilities. Cases of DS children erupting their first molars at 8 years and 9 months *vs*. the age of 6-8 years exhibited by an average rise is an instance of the occurrence of this tendency [7].

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Received: January 01, 2025; Revised: May 16, 2025; Accepted: May 21, 2025
DOI: https://doi.org/10.37184/jlnh.2959-1805.3.24

Craniofacial Malformations

The malformations of the facial middle third are the most common type of complication for people with DS, the negative disorders of this region. Apart from mouth opening, the person with these facial abnormalities will experience a "V"-shape with a high-peak palate, which may further cause an insufficiency in the region. The fact that in the case of 90% of DS, apart from the sinuses of the maxillary, the bones are smaller in shape, as well as distinguishes them from people who do not have this disease.

Dental and Skeletal Anomalies

There is a high probability that some disorders of craniofacial dysplasia will include the incidence of open bite in front of the previous and the orientation of lower incisors toward the front of the mouth. The other oral abnormalities seen to be predisposed in DS patients are complex; they encompass periodontal disease and frequent recurrent respiratory infections that hypoventilate, and thus the patients need to use their mouths for breathing and xerostomia, which is a condition of dry mouth [8].

Enamel Defects and Immune Complications

In the same way, fusion, twinning, clastogenicity, as well as an abnormal shortening of the roots are all known issues in enamel that can cause mechanical problems as well [8-10]. Among the typical issues is the status of

  ISSN: 2960-2963 (Online) *Journal of Liaquat National Hospital 2025; 3(3): 246-251*

periodontal disease and in the future advanced forms of people to romantic disease in the hosts compared to the changed immune system of DS childbearing women. This is, therefore, a big challenge, which confuses their ability to be competent enough to eliminate organisms harboured in dental plaque. One more thing the section illuminates is the causal association between the two diseases and immune deficiency. The association of the growth of microorganisms on the tooth surfaces with demineralisation manifests as cavities [11, 12].

Developmental Delay and Midfacial Growth Issues

Intellectual disability, which is universal to the patient, presents the pregnant sign of this syndrome. DS occurrence is strongly correlated with age, and the older mother has a higher possibility of acquaintance. Usually observed in DS is the fact that the midface bone does not completely develop, causing the base of the palate to be short from front to back. Most bite problems relate to the vertical or horizontal misalignment of jaws, which may result in anterior open bite, posterior crossbite, and the reduction of the upper dental arch width. Such functional degradation in oral functions like chewing, swallowing, and speech can be brought out by those bite irregularities. A DS child has a very high rate of these occlusion abnormalities. They should always be a major concern in the treatment plan since they have a very significant impact on the entire person's overall well-being [13-17].

Hypodontia in Down Syndrome

Hypodontia in individuals with Down syndrome (DS) as a whole is similar not only regarding the kind of missing teeth but also the localisation to the general population, but is observed to have a greater occurrence in DS individuals. Most commonly, hypodontia in DS is unilateral when people are disproved to the instincts of the genetic similarity from both sides of the body. However, specific symmetry patterns in crooked-toothed children with DS have not been thoroughly described yet. Whether genetics, environmental factors, or the way they interact, with hypodontia, all are complex factors that influence its aetiology, as well as the process of its development [3, 18-24].

Orthodontic Treatment Considerations

The orthodontic treatment of people with DS typically requires highly coordinated care that brings together multiple medical fields, and they often encounter some major difficulties, especially cooperation deficiencies, as they often have mental illnesses. Consequently, the Dental Aesthetic Index (DAI) can perform the dual function of an indicator for orthodontists to determine those in need *versus* those who can just wait, as well as an educator to both parents and patients. The biggest limiting factor for treatment success, therefore, depends on a jointly held and mutual understanding of treatment goals among the clinician and the parents [18-20].

Comprehensive Care and Systemic Involvement

The conditions associated with the syndrome of Down undoubtedly make the patient's range of social and educational life equally suffering, the main cause being the developmental delay. These problems can develop as both acute dilemmas and long-term problems, which eventually lead to the abnormal working of the organism and the development of multiple organ distress [25].

Need for Multidisciplinary Approach

Orthodontists, periodontists, endodontists, prosthodontists, and arthropods should not only be skilled in their fields but also have a multidisciplinary view. One more important thing in comprehensive health care for patients is the need for integrated care, which is essential for ensuring the overall well-being of these patients [26].

Summary

This review explores the relationship between DS and dental anomalies, with a focus on tooth morphology, number, eruption patterns, and structural defects. It seeks to summarise existing studies, point out knowledge gaps, and emphasise therapeutic implications for oral healthcare.

SEARCH STRATEGY

Fig. (1) shows the study selection process for a review. Out of 7,324 records identified from Google Scholar and PubMed, 1,090 remained after removing duplicates. After screening and eligibility assessment, 40 studies were included in the final analysis.

DISCUSSION

A Research in 2018 revealed findings based on 36 parents of DS children, who are members of POTADS (Persatuan Orang Tua Anak dengan Down Syndrome/ Association of Parents with Down Syndrome Children) in Bandung, Indonesia, by a research group team. The study results indicated that among the parents whose children have DS, their knowledge of the child's current oral and dental health had improved significantly more after borrowing (4.44 ± 1.362) and (7.47 ± 2.274) books for their children, and raising the values to (6.69 ± 0.856) [27]. Parents are certainly informed about dental and oral health aspects of their special children as a result of this book on people with DS, given that they were not very knowledgeable about the issue some time back. In that way, they trained themselves to be specialists on the

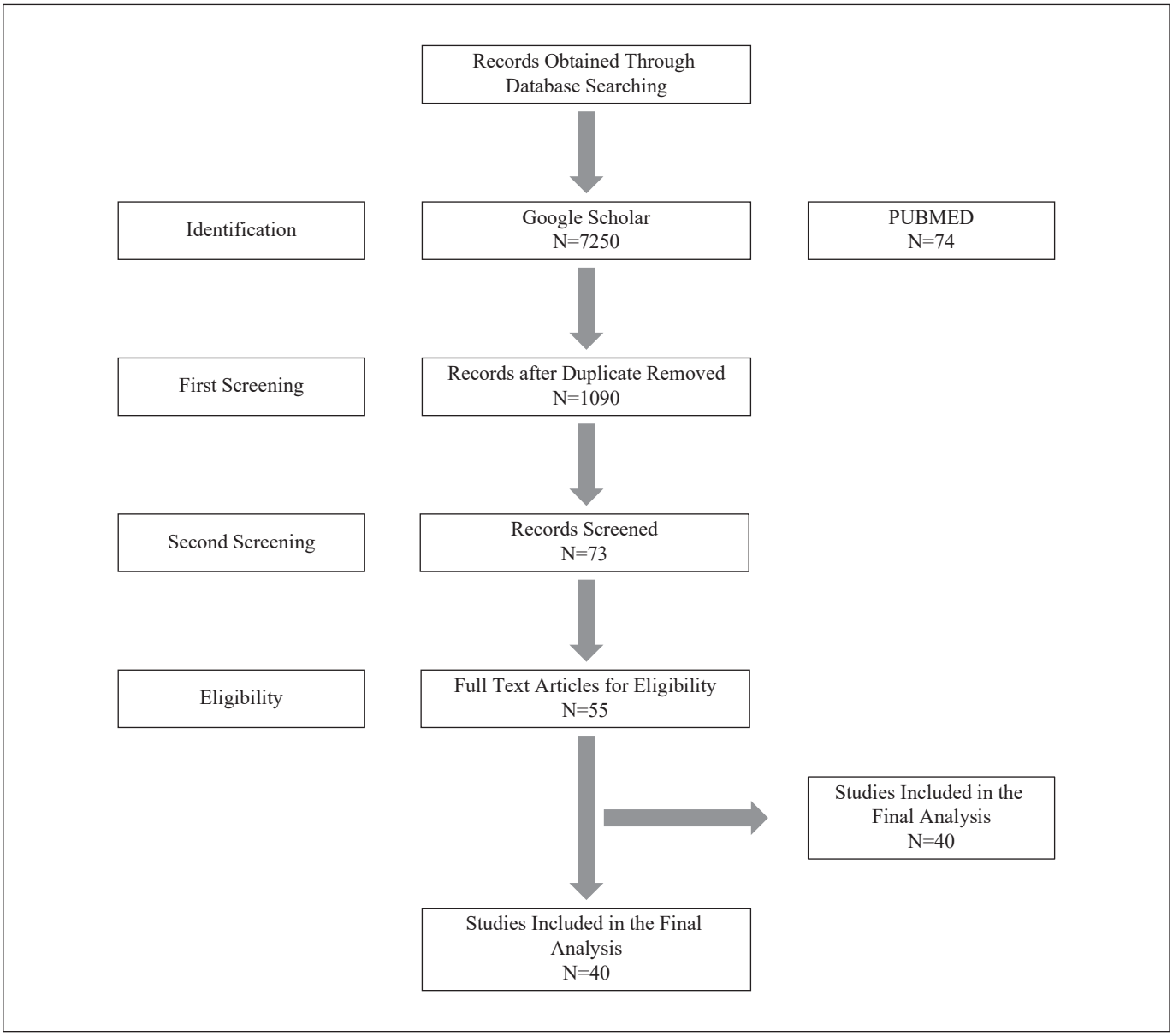

Fig. (1): Flowchart showing the search strategy using Google Scholar and Pubmed.

subject of their children's oral hygiene and in dealing with the challenges they faced.

While a multiple error population study of 2014-2015 made in Portugal and included a methodical approach to the relations between the sister and the brother in the entire sample, it becomes evident that using more than one diagnostic style would be more than enough for the purposes. This study involved the participation of 135 children who were diagnosed with DS, comprising 80 of their siblings of the same age range, 2 to 26. As a result of the study, children of DS and 72% of their siblings had Angle first class equal to approximately 76% and 72% respectively. The frontal plane projections of the control group exhibited more frequent occlusal irregularities, which are, for example, the notch at the lateral edge of the posterior arch($P<0.001$), the open bite anterior ($P=0.005$), and the crossbite anterior ($P=0.001$). The category of "siblings" appears to conduct "translate" significantly (P 0.08). In contrast to their siblings, children with DS have a high chance of having the anterior open bite, the posterior crossbite, and both DS [13].

In another study in Saudi Arabia, 100 Saudi people with DS from four to 14 years of age (group I) underwent oral and dental malformation screening through the method of intraoral examination. The second group of healthy children (group II) with the same age range are taken from the Saudi population and they will be used as a control group. DS patients combined had one or more types of oral and dental issues, with macroglossia (94%) being the most common, followed by narrow palatal vault and fissured tongue, which demonstrated economic

conditions by showing 76% more and 74% more cases, respectively [6].

In the 2019 seminal work by Luly Anggraini, where Ike Sitis Indiarti and Mochamad Fahlevi Rizal were the co-authors, the accessible age of individuals aged 14-53 years old were involved in the study for a total of 274 individuals. Women took up 70 (40.2%) *versus* male counterparts 104 (59.8%), with age at a modal rate of 19.2 years. Based on the fact that participants' names indicated dental abnormalities by no fewer than 81 cases of hypodontia, 12 cases of supernumerary teeth, and both these anomalies occurring concurrently, the researchers concluded that adolescents in the obese and overweight groups were more likely to have dental abnormalities [28].

Besides, a new article in 2011 by Dr. Sanjay Suri explored 25 patient profiles that had no bias for their eye colours. It was concluded in the study that the third molars caused missing teeth to be absent, and thus, in one study, it was 92% and in another, it was 56%. The findings of the study showed that the prevalence of hypodontia was greater in the female siblings and that the most common form of the disorder was the severe one. The topmost frequency of magnetic teeth appeared predominantly in molars of both the maxillary and mandibular sides, including third molars, mandibular lateral incisors, mandibular second premolars, and mandibular incisors. The frequency found at the bottom was the maxillary second premolars, maxillary second molars, and mandibular lateral incisors. More interesting was the occurrence of this 50% of the sample, since it had both third molars missing as well [29].

A study of a cohort of infants born in 2002, who were part of a nationwide prospective study, investigated possible links among upper airway function, hearing, dentition, and craniofacial features in children with Down syndrome. Among the 26 children who were followed up, hypodontia was observed in the permanent dentition (excluding third molars) in 61.5%. Of these, 76.2% exhibited hypodontia affecting two or more permanent teeth. Four children were diagnosed with an extremely rare condition known as oligodontia. The teeth most frequently missing were the maxillary lateral incisors, followed by the mandibular and maxillary second premolars. Most cases of hypodontia were bilateral, accounting for approximately 68.9% of affected individuals. A considerable number of children with Down syndrome in the study also exhibited teeth showing root anomalies suggestive of permanent ankylosis. Compared with the general population, right-sided tooth agenesis in children with Down syndrome occurred almost twice as often as left-sided cases [30–33].

In another study, conducted by Emilia Severin, karyotype examinations revealed three types of chromosomal abnormalities: Triploidy, translocation, and mosaicism involving chromosome 21. These anomalies affect the cell structure. The predominant (most often observed) dental anomaly was hypodontia, which was very frequently noted. Microdontia occurred next. One of the most notable oral manifestations found during the post-mortem examination of the victims was the destruction of all the patient's dental surfaces. Most carriers of 21 trisomy may not have serious consequences for their lives and hence are not considered a factor of high risk [34-39].

CONCLUSION

In conclusion, dental abnormalities such as delayed eruption, hypodontia, malocclusion, and craniofacial underdevelopment are highly prevalent in people with DS. Essential activities, including speech, chewing, and swallowing, are greatly impacted by these oral health issues, which are frequently exacerbated by cognitive and behavioural constraints. As a result, these issues have an influence on people's entire quality of life.

Dental surgery has a good impact on health, including general health, dental health, and other health conditions. This will help to drastically improve the quality of life of patients, which is a task that dentists should commit themselves to, during which they should adopt simple procedures. In most instances, the very common gaps are those of the third molars, the top lateral incisors, and the lower premolars of the second dentition. As such, considering mandibular hypodontia issues without a complex perspective of the patient treatment becomes a portentous act for the patient's well-being.

FUTURE INTENTS

- There is a need for robust, longitudinal research to better understand the etiology and progression of dental anomalies in DS, especially concerning genetic and environmental interactions influencing conditions like hypodontia.
- Educational initiatives targeting parents and caregivers can enhance understanding of oral health needs and improve adherence to treatment plans, ultimately contributing to better long-term outcomes.

FUNDING

None.

CONFLICT OF INTEREST

The authors declare no conflict of interest.

ACKNOWLEDGEMENTS

The authors would like to thank Dr. Asma Shahid and Dr. Saba Shahid for their valuable discussions and support.

The authors would like to thank the reviewers for their helpful feedback on the manuscript.

AUTHORS' CONTRIBUTION

Dr. Aamna Batool: Conceptualised the review topic, performed the literature search.

Dr. Duaa Ali: Selected the sources and wrote the initial draft of the manuscript.

Dr. Abul Faraz: Identify authors, involved in critical revisions and final approval.

Dr. Amna Rehman and Dr. Samreen Malik: Supervised the project and approved the final manuscript version.

Dr. Sadaf Mashood and Dr. Wajida Jawed: Methodology development.

REFERENCES

1. Akhtar F, Bokhari SRA. Down Syndrome. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2020. PMID: 30252272
2. Dyasanoor S, Deverashetty A. Dental management and orodental features of a child with Down's Syndrome. Indian J Dent Adv 2016; 8(2): 119-22. DOI: https://doi.org/10.5866/2016.8.10119
3. Antonarakis SE, Skotko BG, Rafii MS, Strydom A, Pape SE, Bianchi DW, *et al*. Down syndrome. Nat Rev Dis Primers 2020; 6(1): 9. DOI: https://doi.org/10.1038/s41572-019-0143-7 PMID: 32029743
4. Cuoghi OA, Topolski F, de Faria LP, Occhiena CM, Ferreira ND, Ferlin CR, *et al*. Prevalence of dental anomalies in permanent dentition of brazilian individuals with down syndrome. Open Dent J 2016: 10: 469-73. DOI: https://doi.org/10.2174/1874210601610010469 PMID: 27733874
5. Kazemi M, Salehi M, Kheirollahi M. Down syndrome: current status, challenges and future perspectives. Int J Mol Cell Med 2016; 5(3): 125-33. PMID: 27942498
6. Fansa H, Salama RI, Filfilan S. The prevalence of oral and dental anomalies in Down syndrome children in Western region, Saudi Arabia. Int J Health Sci Res. 2019; 9(9): 309-15. DOI: https://doi.org/10.5402/2012/837270
7. Bastos JL, Peres MA, Peres KG, Barros AJD. Infant growth, development and tooth emergence patterns: a longitudinal study from birth to 6 years of age. Arch Oral Biol 2007; 52(6): 598-606. DOI: https://doi.org/10.1016/j.archoralbio.2006.12.001 PMID: 17224130
8. Descamps I, Fernandez C, Van Cleynenbreugel D, Van Hoecke Y, Marks L. Dental care in children with down syndrome: a questionnaire for Belgian dentists. Med Oral Patol Oral Cir Bucal 2019; 24(3): e385-e391. DOI: https://doi.org/10.4317/medoral.22129 PMID: 31011136
9. Singh A, Bhatia HP, Sharma N. Coexistence of fusion and concrescence of primary teeth: in a child with Down syndrome. Spec Care Dentist 2017; 37(3): 147-9. DOI: https://doi.org/10.1111/scd.12218 PMID: 28140477
10. Rohilla M, Rabi T. Etiology of various dental developmental anomalies-Review of literature. J Dent Prob Solut 2017; 4(2): 19-25. DOI: http://doi.org/10.17352/2394-8418.000042
11. Martins M, Mascarenhas P, Evangelista JG, Barahona I, Tavares V. The incidence of dental caries in children with Down syndrome: a systematic review and meta-analysis. Dent J (Basel) 2022; 10(11): 205. DOI: https://doi.org/10.3390/dj10110205 PMID: 36354650
12. Bull MJ, Committee on Genetics. Health supervision for children with Down syndrome. Pediatrics 2011; 128(2): 393-406. DOI: https://doi.org/10.1542/peds.2011-1605 PMID: 21788214
13. de Siqueira Mellara T, Pardini LC, Nelson-Filho P, da Silva RAB, da Silva LA, de Queiroz AM. Occurrence of hypodontia, supernumerary teeth and dental anomalies in Brazilian individuals with Down syndrome. J Disabil Oral Health 2011; 12(1): 31-4.
14. Macho V, Andrade D, Areias C, Coelho A, Melo P. Comparative study of the prevalence of occlusal anomalies in Down Syndrome children and their siblings. J Adv Medicine Med Res 2014; 4(35): DOI: https://doi.org/10.9734/bjmmr/2014/12688
15. Acerbi AG, de Freitas C, de Magalhaes MH. Prevalence of numeric anomalies in the permanent dentition of patients with Down syndrome. Spec Care Dentist 2001; 21(2): 75-8. DOI: https://doi.org/10.1111/j.1754-4505.2001.tb00230.x
16. Moraes ME, Moraes LC, Dotto GN, Dotto PP, Santos LR. Dental anomalies in patients with Down syndrome. Braz Dent J 2007; 18(4): 346-50. DOI: https://doi.org/10.1590/s0103-64402007000400014 PMID: 18278307
17. Mubayrik AB. The dental needs and treatment of patients with Down syndrome. Dent Clin North Am 2016; 60(3): 613-26. DOI: https://doi.org/10.1016/j.cden.2016.02.003 PMID: 27264854
18. Goud ES, Gulati S, Agrawal A, Pani P, Nishant K, Pattnaik SJ, *et al*. Implications of Down's syndrome on oral health status in patients: A prevalence-based study. J Family Med Prim Care 2021; 10(11): 4247-52. DOI: https://doi.org/10.4103/jfmpc.jfmpc_885_21 PMID: 35136797
19. Doriguetto PV, Carrada CF, Scalioni FA, Abreu LG, Devito KL, Paiva SM, *et al*. Malocclusion in children and adolescents with Down syndrome: A systematic review and meta-analysis. Int J Paediatr Dent 2019; 29(4): 524-41. DOI: https://doi.org/10.1111/ipd.12491 PMID: 30834602
20. Mohamed AMFS, Wei TZ, Sean CJ, Rosli TI. Comparison of the malocclusion and orthodontic treatment needs of Down syndrome and non-syndromic subjects by using the dental aesthetics index. Spec Care Dentist 2023; 43(5): 554-60. DOI: https://doi.org/10.1111/scd.12790 PMID: 36269010

21. Sekerci AE, Cantekin K, Aydinbelge M, Ucar Fİ. Prevalence of dental anomalies in the permanent dentition of children with Down syndrome. J Dent Child (Chic) 2014; 81(2): 78-83. PMID: 25198950

22. Abdul Rahim FS, Mohamed AM, Nor MM, Saub R. Malocclusion and orthodontic treatment need evaluated among subjects with Down syndrome using the Dental Aesthetic Index (DAI). Angle Orthod 2014; 84(4): 600-6. DOI: https://doi.org/10.2319/062813-480.1 PMID: 24417495

23. Popoola BO, Onyejaka N, Folayan MO. Prevalence of developmental dental hard-tissue anomalies and association with caries and oral hygiene status of children in Southwestern, Nigeria. BMC Oral Health 2017; 17(1): 8. DOI: https://doi.org/10.1186/s12903-016-0236-6 PMID: 27430583

24. Nirmala SV, Saikrishna D. Dental concerns of children with Down's syndrome–an overview. J Pediatr Neonatal Care 2017; 6(3): 00248. DOI: https://doi.org/10.15406/jpnc.2017.06.00248

25. Rahimi T, Khazir Z. Perceived experiences of life problems for parents with a Down syndrome child. Health Edu Health Promot 2019; 7(3): 147-54. DOI: http://dx.doi.org/10.29252/HEHP.7.3.147

26. de Moraes Pini D, Fröhlich PCGR, Rigo L. Oral health evaluation in special needs individuals. Einstein (Sao Paulo) 2016; 14(4): 501-7. DOI: https://doi.org/10.1590/s1679-45082016ao3712 PMID: 28076597

27. Putri MH, Koesoemah HA, Widyastuti T. The effect of using dental and oral health book on the knowledge and skills of parents with Down Syndrome children. Padjadjaran J Dent 2018; 30(3): 233-42. DOI: https://doi.org/10.24198/pjd.vol30no3.19277

28. Anggraini L, Rizal MF, Indiarti IS. Prevalence of dental anomalies in Indonesian individuals with down syndrome. Pesquisa Brasileira em Odontopediatria e Clínica Integrada 2019; 19: e5332. DOI: http://doi.org/10.4034/PBOCI.2019.191.147

29. Suri S, Tompson BD, Atenafu E. Prevalence and patterns of permanent tooth agenesis in Down syndrome and their association with craniofacial morphology. Angle Orthod 2011; 81(2): 260-9. DOI: https://doi.org/10.2319/070910-391.1 PMID: 21208078

30. Andersson EM, Axelsson S, Austeng ME, Øverland B, Valen IE, Jensen TA, *et al*. Bilateral hypodontia is more common than unilateral hypodontia in children with Down syndrome: a prospective population-based study. Eur J Orthodont 2014; 36(4): 414-8. DOI: https://doi.org/10.1093/ejo/cjt063 PMID: 24014738

31. Esperanza RA, Evans A, Tucker D, Paranjothy S, Hurt L. Hospital admissions in infants with Down syndrome: a record-linked population-based cohort study in Wales. J Intellect Disabil Res 2022; 66(3): 225-39. DOI: https://doi.org/10.1111/jir.12903 PMID: 34859911

32. Ghaith B, Kowash MB, El-Halabi M. Dental implications of Down Syndrome (DS): review of the oral and dental characteristics. Appl Clin Res Clin Trials Regulat Aff 2016; 3(3): 151-8 DOI: https://doi.org/10.2174/2213476x03666160901125732

33. Shore S, Lightfoot T, Ansell P. Oral disease in children with Down syndrome: causes and prevention. Community Pract 2010; 83(2): 18-21. PMID: 20222361

34. Severin E, Paun A, Baltag R, Stan A, Funieru C. Common, rare, and individual oro-dental findings in people with Down syndrome. J Int Oral Health 2016; 8(10): 964-8.

35. Antonarakis SE, Skotko BG, Rafii MS, Strydom A, Pape SE, Bianchi DW, *et al*. Down syndrome. Nat Rev Dis Primers 2020; 6(1): 9. DOI: https://doi.org/10.1038/s41572-019-0143-7 PMID: 32029743

36. Bull MJ. Down syndrome. N Engl J Med 2020; 382(24): 2344-52. DOI: https://doi.org/10.1056/nejmra1706537 PMID: 32521135

37. Desai SS. Down syndrome: a review of the literature. Oral Surg Oral Med Oral Pathol Oral Radiol Endod 1997; 84(3): 279-85. DOI: https://doi.org/10.1016/S1079-2104%2897%2990343-7 PMID: 9377191

38. Hendrix JA, Amon A, Abbeduto L, Agiovlasitis S, Alsaied T, Anderson HA, *et al*. Opportunities, barriers, and recommendations in Down syndrome research. Transl Sci Rare Dis 2021; 5(3-4): 99-129. DOI: https://doi.org/10.3233/trd-200090 PMID: 34268067

39. AlShahrani I, Togoo RA, AlQarni MA. A review of hypodontia: classification, prevalence, etiology, associated anomalies, clinical implications and treatment options. World J Dent 2013; 4(2): 117-25. DOI: https://doi.org/10.5005/jp-journals-10015-1216