Co-morbidity of down syndrome with autism spectrum disorder: Dental implications

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ABSTRACT

Over the past several years, numerous studies have emerged documenting the high incidence (1–11%) of comorbidity of autism spectrum disorders (ASD) in Down syndrome (DS). While children with these health issues are reported to be more cognitively impaired presenting significantly lower IQ scores, they also demonstrate differences in social and expressive language skills when compared to their coequals with DS only. More than that subjects with DS and ASD comorbidity exhibit atypical behaviour manifested by stereotypic anxiety and social withdrawal when compared to DS alone.

This article provides a brief understanding of this challenging concurrence along with a case report of a 12-year-old male patient with ASD-DS condition reporting with multiple missing teeth (Oligodontia).

1. Introduction

Over the past several years, numerous studies have emerged documenting the high incidence (1–11%) of comorbidity of Autism Spectrum Disorders (ASD) in Down Syndrome^{1,2} suggesting an implication for a shared genetic etiology.³ While children with DS-ASD-comorbidity are reported to be more cognitively impaired presenting significantly lower IQ scores, they also demonstrate differences in social and expressive language skills when compared to their coequals with DS only. Children with DS-ASD have been described to show an increased frequency of developmental regression along with the greater potential for neurological afflictions such as seizures, dysfunctional swallowing, severe hypotonia, and weaker motor skill, congenital cardiac defects, gastrointestinal tract aberrance, ophthalmological disorders, pneumonia, and sleep disturbances.^{2,4,5} More than that subjects with DS and ASD comorbidity also exhibit atypical behaviour along with a stereotypic anxiety and social withdrawal when compared to DS alone.⁶

This article provides a brief understanding of this challenging concurrence along with a case report of a 12-year-old male patient with ASD- DS condition reporting with multiple missing teeth (Oligodontia).

1.1. Case report

A 12-year boy reported to the Department for Pediatric and

Preventive Dentistry, Faculty of Dentistry, JMI with a chief complaint of acute toothache and mobility associated with multiple teeth. The patient was of short stature and mentally challenged with the typical appearance of Down syndrome viz the saddle nose deformity, hypertelorism, midface hypoplasia with the retruded maxilla and protruded mandible including the intra-oral aspects of the high arch palate and macroglossia of the tongue (Fig. 1). The child also displayed signs of hyperactivity, lack of attention to commands and limited speech. There was no history of seizures and no evidence of infantile spasm and abnormal oral habit in the patient. Parent's medical history was non-contributory. At the age of 8 years, the child was referred to a premium tertiary centre because of unusual social progress, poor language development, and violent behaviour. The concerns about his hearing were also expressed because he appeared to neglect instructions, but hearing tests were found to be normal. The patient's TSH values were found to be high (6.14 mU/L) so 25 mg Thyronorm was prescribed for 6 months. At present, the child was not under any medications. On the VSMS scale, (Vineland Social Maturity Scale) a substitute to an IQ test in cases where the child is unresponsive to the IQ test, the patient was diagnosed with a moderate intelligence disability (SA-54, CA-135, SQ-40). On further investigations, the child fulfilled the criteria of Autism Spectrum Disorder based on the Child Behaviour Checklist (CBCL) scale.

On intraoral examination grade III mobility was found in 11, 21, 34, 75, 84 and generalized pathological spacing in dentition. OPG

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Fig. 1. Typical appearance of Down syndrome.

Fig. 2. OPG radiograph showing multiple missing teeth and bilateral transposition of 13, 14 and 23, 24.

radiograph revealed the absence of 12, 22, 31, 35, 37, 41, 42, 45 teeth. There was also presence of transposition involving 23, 24 and 13, 14. Based on radiographic evaluation a dental diagnosis of Oligodontia with bilateral maxillary canine/first premolar transposition and chronic periodontitis was made (Fig. 2).

Since the child was uncooperative in behaviour dental treatment was not possible in a regular dental setup. Parents were instructed for regular dental check-ups and also about maintenance of oral hygiene.

2. Discussion

The chances of dental anomalies in subjects with Down syndrome are quite high, with 15% prevalence of maxillary canine/first premolar transposition⁷ and 63% cases of oligodontia⁸ reported in the dental literature. Many studies have also indicated that the reduced ability to maintain adequate plaque control and altered immune responses, especially among the younger age group of DS, elevates the risks of

periodontitis in juveniles by 30–40%.⁸ This present case also presented with oligodontia and bilateral transposition of the maxillary canines with the first premolars with severe mobility and generalized bone loss resulting in acute pain and sensitivity. It is also common for children with DS-ASD comorbidity to have malocclusions like anterior open bite, dental crowding or spacing due to the presence of abnormal oral habits of bruxism, tongue thrusting or lip biting.⁹

Dental management of a child or adolescent with only Down syndrome does not cause many obstacles, because they are mostly pleasant and well behaved. However, in patients with DS-ASD comorbidity, the reduced cognitive functioning, sensory hypersensitivities (visual, auditory, olfactory or gustatory stimuli)¹⁰ and limited motor skills make the oral care attempt more problematic. The subject can get annoyed even by a light touch or by a non-invasive dental procedure and they may fall back during the dental examination or display violent behaviour, like headbanging and temper tantrums^{10,11} In this case, too, the parents felt that the child had frequent mood swings and got angry without provocation.

Usually, patients with mild MR and lacking behavioural problems are likely to be cooperative during dental treatment. The presence of parents or assistants during the treatment can enhance their positive conduct. The desired behaviour may also be achieved by behaviour shaping and desensitization techniques,¹⁰ such as the tell-show-do technique and by giving short clear verbal commands along with positive and negative verbal reinforcements. Mouth props can be used to keep the mouth open during the various dental procedures however use of hand over mouth exercise and physical restraints are not suggested.^{10,12} It is also recommended that the immobilization of the patient should be followed for the protection of the dental team and not for the convenience during dental treatment.¹¹ Our patient's compliance was very poor and was unable to follow any instructions on the dental chair and hence the treatment was deferred but parents were motivated for oral hygiene maintenance.

Besides, the patients with ASD are usually on anticonvulsants agents, eg carbamazepine and valproate, which can cause, leukopenia, thrombocytopenia, and decreased fibrinogen activity. The use of these medications, combined with NSAIDs or any analgesics may impair the haemostatic mechanism and result in bleeding. Any surgical therapy should be planned after thorough blood investigations. Such patients are also on Methylphenidate (CNS stimulant) to control their hyperactivity; hence chances of inadvertent hypertensive mishaps are more if local anaesthesia is administered in excess or intravascularly. The use of antibiotics like erythromycin and clarithromycin may cause carbamazepine toxicity by inhibiting its metabolism in the liver.^{12,13} More frequently, these patients display atypical and inappropriate reactions, with oral and IV sedation^{12,13} hence in 40% of cases, dental treatment is possible under general anaesthesia only.¹⁰

Non-verbal, Symbolic Expression or Visual pedagogy has been proven to be an efficient technique in introducing the basic steps of dentistry and tooth brushing by series of coloured pictures.¹⁰ During each dental visit, the treating dentist should document every approach in dealing with such individuals in the patient's record file and should note down any characteristic preferences such as music, flavours, comforting words, gestures or objects that helped in the successful accomplishment of the treatment.¹¹

3. Conclusion

The dental treatment of patients with DS- ASD comorbidity can be challenging because of their behavioural, cognitive, and social restraints as compared to those with DS in isolation. Based on the patient's age, cooperation and extent of dental treatment, the use of sedation and general anaesthesia can be considered for the dental management.

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