CASE REPORT

Oral Manifestations and Dental Management in Emanuel Syndrome: A Case Report

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ABSTRACT

Oral Manifestations and Dental Management in Emanuel Syndrome: A Case Report

Emanuel syndrome (ES, OMIM 609029), also known as supernumerary der(22)t(11;22), is a rare condition (1:110000 people) characterized by pre- and postnatal developmental delays and multiple congenital abnormalities. This is a case report of an 8-year-old boy with ES who has cardiac, musculoskeletal, auditory, visual, and renal problems and a general growth restriction with an intellectual disability and impaired speech. All primary dentition demonstrated hypomineralized areas, deep caries and mild gingival hyperplasia. Delayed eruption of both primary and permanent dentition, congenitally missing permanent second premolars and primary lateral incisors, short root forms and open apices of permanent molars, and a right posterior cross-bite were significant oro-dental findings. A necessary pulpotomy and subsequent composite restorations and extractions were performed under general anaesthesia. This case report provides a comprehensive list of systemic and oro-dental ES findings along with late dental interventions and 2-year follow-up observations that have not been reported in detail previously.

KEYWORDS

Emanuel Syndrome; Hypodontia; Hypomineralization; Micrognathia; Cleft Lip/Palate

ÖZ

Emanuel Sendromunda Oral bulgular ve Dental Tedaviler

Emanuel sendromu (ES, OMIM 609029), süpernümerer der(22)t(11;22) olarak da bilinen, doğum öncesi ve sonrası gelişimsel gecikmeler ve çoklu konjenital anormallikler ile karakterize nadir bir sendromdur (1:110000 kişi). Bu, kardiyak, kas-iskelet sistemi, işitsel, görme, böbrek problemleri, zihinsel engeli, konuşma bozukluğu ve genel bir büyüme kısıtlaması olan ES'li 8 yaşındaki bir çocuğun vaka raporudur. Tüm primer dentisyonda hipomineralize alanlar, derin çürükler ve hafif diş eti hiperplazisi gözlendi. Hem primer hem de kalıcı dişlenmede sürmede gecikme, konjenital eksik kalıcı ikinci premolarlar ve primer lateral kesici dişler, kısa kök formları ve kalıcı azı dişlerde açık kök ucu ve sağ arka bölgede çapraz kapanış önemli oro-dental bulgulardı. Genel anestezi altında gerekli pulpotomi işlemlerini takiben kompozit restorasyonlar ve ekstraksiyonlar yapıldı. Bu olgu sunumunda, geç diş müdahaleleri ve daha önce ayrıntılı olarak bildirilmemiş 2 yıllık takip gözlemleri ile birlikte sistemik ve oro-dental ES bulgularının kapsamlı bir listesi sunulmaktadır.

ANAHTAR KELİMELER

Emanuel Sendromu; Hipodonti; Hipomineralizasyon; Mikrognati; Yarık Dudak/Damak

INTRODUCTION

Emanuel syndrome (ES, OMIM 609029), also known as supernumerary der(22)t(11;22) syndrome, is characterized by pre- and postnatal growth restrictions and multiple congenital abnormalities such as microcephaly, hypotonia, mental retardation, ear anomalies, preauricular tags or pits, a cleft or high-arched palate, congenital heart defects, kidney abnormalities and genital abnormalities in males.[1] In >99% of patients with ES, one of the parents is a balanced carrier of a t(11;22)(q23;q11.2) translocation, most often the mother, and has a normal phenotype; it is a rare disorder with a reported prevalence of 1 in 110000 people.[2] It should be taken into consideration that the existing literature contains limited information about oral and dental findings and dental management in ES.

CASE REPORT

Our male patient with ES, age 8, was referred for dental evaluation at Trakya University (T.U.), Faculty of Dentistry, Department of Pediatric Dentistry by his paediatrician. The patient's detailed medical history and dental findings are documented in Table 1. It was the patient's first dental visit. He was uncooperative during the examination and routine dental radiography in the clinical setting. His mother's chief complaint was pain, and she could not provide oral hygiene measures properly (not brushing daily/flossing). Dental treatments were performed on the patient under general anaesthesia (GA) immediately. Necessary dental radiographs and photographs were taken under GA before dental treatments. A mobile upper right central incisor (FDI #61) was already knocked out due to nasal intubation using a laryngoscope. Consent for publication was provided by the patient's parents before drafting this case report.

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Table 1.

Medical and Dental Findings

Prenatal History	The first and only pregnancy (no history of abortion or miscarriage) of a 5-year married couple without a consanguineous marriage. Maternal age at conception was 22 years. Uneventful prenatal period.
Birth	Full-term by vaginal delivery. Birth weight 3.25 kg and length 52 cm.
Genetic Findings	47,XY,+der(22)t(11;22) (q23.3; q11.2) (unbalanced translocation) The mother was found to be a balanced carrier: 46,XX, t(11;22)(q23.3;q11.2)
Medications	Dorzolamide/Timolol 2%+0.5% eye drops (1 drop per eye-BD)
Adjunct Therapies	Special education both at school and home with one-on-one support
Vitals at Age 8	Weight (25 kg), height (128 cm). BMI was 15.26 (normal), and blood pressure was 90/60 mm of Hg.
Craniofacial	Preauricular tags and pits, enlarged lateral ventricles, short neck, low posterior hairline, and hooked nose with low-set columella
Oro-dental	Angular lip pits Delayed eruption of both dentition (primary and permanent, 2-3 years) Cleft-palate at birth (surgically corrected) Posterior cross-bite due to transverse maxillary deficiency Open bite Tulip-shaped primary lateral and permanent central incisors Hypodontia (missing permanent second premolar) Morphological abnormalities in crowns of lower permanent molars Short-root anomaly Macroglossia
Auditory	Recurrent otitis media Bilateral hearing loss (using hearing aid)
Ophthalmic	Hypertelorism Right eyelid ptosis Glaucoma
Central Nervous System	Delayed psychomotor development Intellectual disability Impaired speech (non-verbal) Hypotonia
Immune	Congenital immunological deficiency
Cardiovascular	11 mm secundum atrial septal defect Patient's paediatric cardiologist did not require antibiotic prophylaxis for dental care.
Genitourinary	Undescended testicle and inguinal hernia (surgically corrected at age 7) Renal defects
Gastro-intestinal	Gastroesophageal reflex Feeding difficulties (spoon feeding) No sphincteric control (wearing diaper)
Musculoskeletal	Delayed skeletal age (2-3 years) Dependent mobility (using stroller) Scoliosis Kyphosis

Intraoral Evaluation: Soft Tissues

The patient generally showed mild gingival hyperplasia (which is difficult to say it is due to ES since the patient had poor oral hygiene), a shallow V-shaped palate with a scar on the midline indicating a surgically corrected Class II cleft in Veau classification and macroglossia (Figure 1). The patient had Mallampati grade III airway and grade II tonsillar scores. Other oropharyngeal soft tissues were normal.

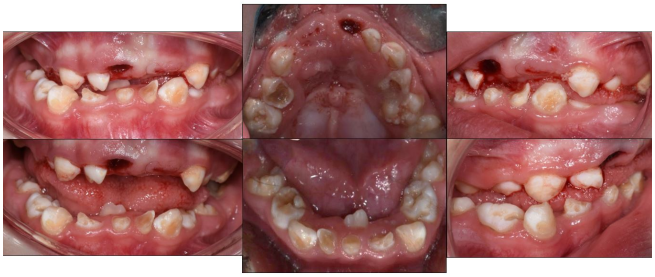

Figure 1.

Intraoral pictures before dental treatment. Intraoral centre (A, D), intraoral maxillary occlusal (B), intraoral left (C), intraoral mandibular occlusal (E), and intraoral right (F) aspect. Teeth present: #55, #54, #53, #52, #63, #64, #65, #75, #74, #73, #71, #41, #81, #82, #83, #84, and #85

Intraoral Evaluation: Hard Tissues

The patient was in early mixed dentition with one permanent lower incisor, and the permanent first molars erupted (Figure 1). Mandibular primary lateral and central incisors were mobile, and permanent central incisors erupted lingually. Tooth #61 was exfoliated at the age of 7 and teeth #62 and #82 were congenitally missing. All primary dentition demonstrated hypomineralization and deep caries clinically and radiographically in both occlusal surfaces of the primary molars and the buccal surfaces of incisors mostly because of feeding practices (spoon feeding) and poor oral hygiene habits. Radiographic evidence of morphological abnormalities in the mandibular first permanent molars, especially tooth #46, suggested hypomineralization or hypoplasia (Figure 2).

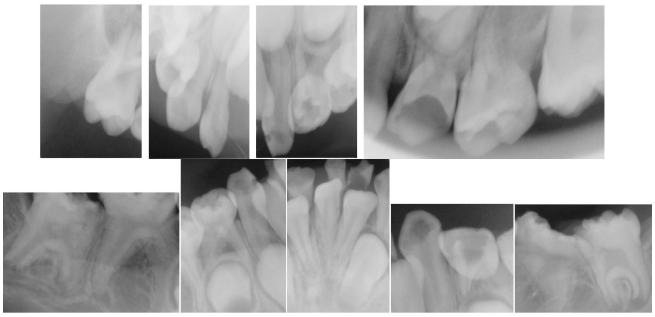

Figure 2.

Periapical radiographs at 8 years of age. Tulip-shaped crowns of erupted primary incisors, deep carious lesions in all dentition, morphological abnormality in #46, short roots and open apices in primary second molars and permanent first molars, congenitally missing permanent second premolars.

Open apices and short root forms in permanent first molars were considered a general developmental delay and noted. A transverse maxillary deficiency-mediated right posterior cross-bite was noted. The mother reported no history of dental trauma.

Dental Treatment

Appropriate dental treatment was managed under general anaesthesia. Teeth #54, #64 and #74 were treated with pulpotomy and restored with composite resin consecutively. After the removal of caries from teeth #55, #53, #52, #63, #65, #75, #73, #83, #84, and #83, restorations were completed with composite resin. Teeth #82, #81, and #71 were extracted (Figure 3).

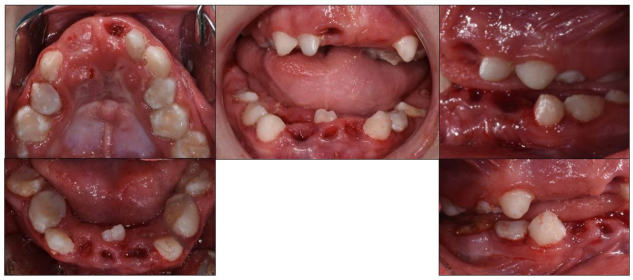

Figure 3.

Intraoral pictures after dental treatment. Intraoral centre (B), intraoral maxillary occlusal (A), intraoral left (C), intraoral mandibular occlusal (D), and intraoral right (E) aspect. Teeth present: #55, #54, #53, #52, #63, #64, #65, #75, #74, #73, #41, #83, #84, and #85

Within the first week of treatment, the patient's oral hygiene was fair, and his periodontal health was better. After two years, his permanent upper and lower incisors and first permanent molars were erupted, and the restorations remained clinically acceptable.

DISCUSSION

Follow-up appointments continue 2 years after managing the dental treatment of this case. The photographs and periapical radiographs presented above are the best available. The patient's limited mouth opening and desire to avoid overexposure to radiation hindered the acquisition of proper visual records. Parental awareness and early intervention in oral health should be the main goal from the standpoint of paediatric dentistry.

Clinical findings such as facial asymmetry, preauricular ear pits, ptosis, cleft palate, crowded teeth, congenitally missing teeth, cardiovascular and genitourinary defects, and musculoskeletal disorders were common.[3] Although the exact mortality rate in ES is unknown, if the patient survives infancy, long-term survival is possible.[4] This demonstrates the need for a multidisciplinary team approach involving paediatric dentists, paediatricians, ophthalmologists, ear, nose and throat (ENT) surgeons, plastic surgeons, geneticists, cardiologists, urologists, gastrologists, speech therapists and special education teachers.

Neurological involvement in ES plays a major role in the duties of the primary caregivers of the patient regarding dental hygiene and feeding practices. Preserving the arch length and vertical dimension become more important in ES as hypodontia and cleft palate are predisposing factors for malocclusion or crowding.

Clinical features in ES, such as hypotonia, intellectual disability, and impaired speech, impact dental treatment delivery, such as the choice of hospital-based GA. Cardiac anomalies have been reported in 57% of ES cases [5], necessitating consultation with a cardiologist to determine the need for antibiotic prophylaxis before dental treatment. Intubation for GA in patients with ES can be challenging due to craniofacial and oropharyngeal abnormalities.

Being a rare syndrome, and to the best of our knowledge, there is only one other relevant case report [3], any information and management experiences especially about the detailed oral and dental problems of patients with ES are a valuable contribution to the ES literature.

CONCLUSION

This is the first case report about dental management in ES. Patients with ES should be referred to paediatric dental clinics to be informed about oral and dental challenges before it is too late. The information obtained during the dental management of this case provides valuable guidance for paediatric dentists regarding safe and effective treatments of patients with ES.

Competing interests:

The authors declare that they have no competing interest.

Financial Disclosure:

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