Dental and Medical Considerations for Patient with Neurofibromatosis Type 1

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Abstract

Neurofibromatosis type 1 (NF1) is an autosomal dominant genetic disorder caused by pathogenic variants in the NF1 gene with an estimated incidence of 1 in 3,000 births. The condition is characterized by café-au-lait macules, neurofibromas, skeletal dysplasia, and craniofacial abnormalities that may significantly impact oral health, growth, and dental management. This case report describes a 7-year-old male with NF1 presenting with severe facial dysmorphia secondary to an extensive plexiform neurofibroma, necessitating comprehensive dental treatment in the operating room that includes dental extractions, restorative procedures, and non-operative management delivered under general anesthesia. Given the progressive craniofacial involvement of NF1, lifelong multidisciplinary coordination among neurosurgery, plastic surgery, otolaryngology, and orthodontics is essential to guide dental and orthodontic care, optimize function and esthetics, and improve overall quality of life.

Etiology and Diagnosis

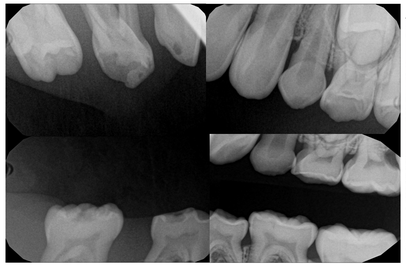
- Pathogenic variants in the NF1 gene on chromosome 17q11.2, which encodes neurofibromin, a tumor suppressor protein that regulates the RAS/MAPK signaling pathway; loss of function results in increased cellular proliferation and tumor formation[3]
- Autosomal dominant inheritance pattern; approximately 50% of cases inherited from an affected parent and 50% arising from de novo mutations[4]
- Diagnosis is primarily clinical, requiring two or more characteristic features[4]
- Molecular genetic testing used in prenatal diagnosis, young children, or atypical cases[4]

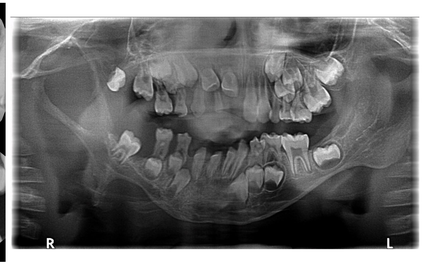
National Institutes of Health (NIH) Diagnostic Features of NF1:[3,4]
Six or more café-au-lait macules (>5 mm in prepubertal children, >15 mm postpubertal)
Axillary or inguinal freckling
Two or more neurofibromas or one plexiform neurofibroma
Optic pathway glioma
Two or more Lisch nodules
Characteristic osseous abnormalities (scoliosis, tibial bowing, and pseudoarthrosis)
Neurologic and cognitive impacts (learning disabilities, attention deficits, and executive function impairments)
First-degree relative with NF1

Dental Considerations

- Intraoral neurofibromas and gingival enlargement[1,5]
- Altered eruption patterns, tooth displacement, impaction, or agenesis, particularly in areas affected by plexiform neurofibromas[1,2]
- Craniofacial growth disturbances: asymmetric jaw growth, enlarged mandibular canals, and malocclusion[1,2]
- Increased risk for gingivitis and plaque accumulation[1]
- Increased risks with surgical procedures due to extent of neural or vascular involvement[1,5]

Case Report





Patient: 7-year-old male presents for routine examination and radiographs in the dental clinic, no chief concern
Medical History: NF1, café-au-lait spots, ankyloglossia, right eye esotropia, congenital glaucoma, parotid mass, plexiform neurofibroma, seizures, speech delay, mixed conductive and sensorineural hearing loss of right ear, facial dysmorphia
Medications/Allergies: none
Social History: lives at home with mother and father
Oral Hygiene: poor, oral hygiene quality is limited by behavior and limited opening; mother attempts brushing twice daily with fluoride toothpaste, but is not flossing
Diet: drinks Boost Kids Essential, milk, water, and juice; snacks on chips, noodles, fruit, and eggs
Caries Risk Assessment: high
Extraoral Exam: exophthalmos of right eye, significant right facial asymmetry extending from forehead to mandible, and restricted opening
Intraoral Exam:
- Caries noted on tooth #B-DO, #C-L, #S-OB, #T-O
- Mobile, over-retained tooth #D
- Deep pits and fissures on tooth #19
- Anterior open bite

Radiographic Finding: hypoplastic appearance of mandible, thin ramus and body of mandible, elongated coronoid process, displacement of #2, agenesis of #31, unerupted #3, #7, #8, #14, and #30

Treatment

Recommended full mouth dental rehabilitation in the operating room under general anesthesia due to the patient's medical complexity, extent of treatment needs, and inability to safely tolerate care in an outpatient setting.

Treatment Plan:
- Stainless steel crowns on #B, #S, and #T
- Composite restorations on #C and #28
- Extraction of #D,
- Sealant on #19
- Dental prophylaxis and fluoride varnish application

Given the diagnosis of NF1and associated craniofacial involvement, special considerations include potential airway challenges due to plexiform neurofibromas, warranting pre-operative anesthesia evaluation and avoidance of in-office sedation due to increased risk of airway compromise and difficulty with emergency management.

Due to patient's high caries risk, increased frequency of fluoride varnish with recall visits is recommended with an emphasis on oral hygiene instruction and nutritional counseling at each visit.

Discussion

Patients with NF1 can present with varying degrees of severity which dictate the type, timing, and extent of treatment needed. These patients benefit from interdisciplinary teams including the following specialties:

- Neurosurgery
- Plastic surgery
- Otolaryngology
- Pediatric Dentistry
- Orthodontics
- Genetics
- Psychology
- Speech
- Nutrition
- Social work

Pediatric dental providers play an important role not only in delivering oral healthcare but also in reinforcing continuity of care by encouraging adherence to recommended medical visits, facilitating referrals, and helping families navigate the healthcare system. Early establishment of a dental home and a team-based approach that emphasizes communication, patient education, and long-term follow-up is essential to optimize both oral and overall health outcomes.

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