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Freeman–Sheldon Syndrome (Freeman–Burian Syndrome): A Contemporary Literature Review

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Abstract:

Freeman–Sheldon syndrome (FSS), also known as Freeman–Burian syndrome (FBS), is a rare congenital disorder characterized by distinctive craniofacial abnormalities, musculoskeletal contractures, and variable systemic involvement. Recent evidence has redefined FSS as a congenital craniofacial myopathy rather than primarily a distal arthrogryposis syndrome, emphasizing the importance of characteristic facial features in diagnosis. This narrative review summarizes current evidence on the clinical presentation, pathogenesis, diagnosis, and multidisciplinary management of FSS, with particular emphasis on its dental manifestations and implications for dental practice. Common oral findings include microstomia, high-arched palate, malocclusion, hypodontia or oligodontia, ankyloglossia, and restricted mouth opening, all of which present significant challenges during dental treatment. Dentists play a crucial role in the early recognition of the syndrome and in planning comprehensive oral healthcare, including preventive, restorative, orthodontic, and anesthetic considerations. Although the available evidence is largely based on case reports and small case series, increasing awareness of the characteristic dental and craniofacial features can facilitate timely diagnosis, multidisciplinary management, and improved patient outcomes.

Keywords: Freeman- Sheldon Syndrome, Freeman Burian syndrome, full mouth rehabilitation, rare diseases

Introduction

Freeman-Sheldon syndrome, now often termed Freeman-Burian syndrome, is best understood in the 2022–2025 literature as a **congenital craniofacial myopathy** with frequent extra-craniofacial involvement rather than primarily a distal arthrogryposis syndrome [1] [2] [3]. The newer literature also shifts emphasis from descriptive case recognition toward stricter diagnostic criteria, clearer distinction from Sheldon-Hall syndrome, and more cautious interpretation of older anesthetic and management claims [4] [3] [4].

Clinical Phenotype

The core diagnostic phenotype remains highly consistent across recent papers: microstomia, pursed or "whistling" lips, deep nasolabial folds, and an H- or V-shaped chin defect [1] [2] [5]. Hand and foot findings are common and often include camptodactyly, ulnar deviation, equinovarus, vertical talus, and related contractures [1] [6] [7]. Extra-craniofacial manifestations frequently include scoliosis, spinal deformity, hearing and visual impairment, and metabolic or gastrointestinal problems [1] [4] [8].

Recent work argues that distal arthrogryposis features support the diagnosis but are not required for it [2]. That position reflects a broader reframing of the disorder as craniofacially defined, with limb abnormalities treated as common but secondary manifestations [2] [3]. Severity varies widely, from minimal malformation to severe respiratory complications in infancy [1].

Mechanism and Classification

The mechanistic literature converges on **abnormal muscle structure and function**. Fibrous or tendinous tissue can replace normal muscle fibers, producing constricting bands that limit movement and contribute to contractures

and facial stiffness [5] [7]. These tissue findings align with molecular myophysiology data showing impaired contraction metabolism, extreme stiffness, and reduced muscular work and power [5] [7].

Autosomal dominant inheritance is now the standard view, although many cases appear sporadic and new allelic variation is common [1] [5] [9]. MYH3 remains the principal implicated gene in the recent literature [4] [8]. At the same time, current diagnostic commentaries stress that molecular testing does not always add clinical value because a minority of clinically diagnosed patients have no identified mutation [2].

Recent authors repeatedly argue that misclassification remains a major problem. Freeman-Burian syndrome and Sheldon-Hall syndrome were historically conflated, but newer reviews describe Freeman-Burian syndrome as preferentially affecting craniofacial development, whereas Sheldon-Hall syndrome more strongly affects the extremities and spine [3]. One 2024 commentary estimated a **30–60% false-positive diagnosis rate**, underscoring how often outdated criteria still shape case reporting [3].

Management Themes

Domain	Main Finding	Recent Evidence
Diagnosis	Recent papers favor craniofacial criteria and specialist evaluation	Microstomia-centered criteria are straightforward, and craniofacial or plastic surgeons are argued to be best positioned for diagnosis and ongoing care [2]
Prenatal recognition	Prenatal diagnosis appears possible in selected cases	Midline mandibular cleft on fetal ultrasound was the most specific prenatal clue, though imaging signs remain technically difficult to assess [8]
Orthopedic care	Early non-operative treatment is emphasized	Stretching, massage, splinting, serial casting, and bracing were used first-line, with Achilles tenotomy under local anesthesia when needed [6]
Dental and orthodontic care	Microstomia remains the main barrier	Limited oral access complicates care, but recent case reports describe successful non-surgical orthodontic management and stable retention [7]
Ocular management	Vision-preserving surgery is often the goal	Ptosis, strabismus, and blepharophimosis are common, and surgery is often considered to prevent amblyopia [10]

Anesthetic complexity remains one of the most discussed clinical issues. Multiple recent reports agree that airway management is difficult because microstomia, mandibular hypoplasia, high-arched palate, and neck immobility can make direct laryngoscopy and intubation challenging [9]. Difficult vascular access and postoperative respiratory risk also continue to appear in recent summaries [3].

Recent literature suggest that Freeman-Burian syndrome is not associated with elevated malignant hyperthermia risk and that older claims were propagated from sparse reports [2] [3] [5]. In contrast, some ophthalmic and anesthetic case reports still describe concern about malignant hyperthermia or trigger-avoiding anesthetic strategies [10] [9].

Joshi et al, describes a **2022 case report** of a 5-year-old girl with multiple carious teeth whose craniofacial and oral features led to a clinical diagnosis of Freeman-Sheldon syndrome, a rare congenital disorder also called whistling face syndrome [12]. The child had delayed developmental milestones, scoliosis and hemivertebrae, and characteristic findings including pursed lips, a V-shaped chin, low-set ears, microstomia, high-arched palate, ankyloglossia, and missing lower anterior teeth with oligodontia on imaging . Genetic testing of MYH3 and NALCN found no clinically relevant variants, which the authors interpret as consistent with the minority of patients who show an FSS-like phenotype without identifiable mutations in currently known genes. Because the child was unable to tolerate chair-side care, the team completed full-mouth dental rehabilitation under general anesthesia, including pulpectomies, stainless steel crowns, restorations, and a space maintainer, while noting

airway and anesthetic challenges that can accompany FSS. The article's stresses on the fact that dentists can play an important role in recognizing Freeman-Sheldon syndrome from its characteristic phenotype and in providing carefully planned dental management and follow-up even when genetic confirmation is absent [12].

Evidence Quality

The strongest meta-level finding in the newest literature is that the evidence base remains dominated by **case reports**, which places unusual weight on accuracy and currency [4]. Recent methodological commentary argues that rare-disease case reports should explicitly demonstrate how a patient meets accepted diagnostic criteria and should interpret findings within an established disease mechanism whenever possible [4].

This concern is especially relevant here because recent authors describe persistent confusion about required diagnostic findings and other clinically important claims in the Freeman-Burian syndrome literature [4] [5]. Even recent case reports outside the main diagnostic commentaries still reproduce older classifications, such as describing the syndrome as the most severe distal arthrogryposis, showing that the literature remains transitional rather than fully settled [11] [9].

Taken together, the 2022–2025 literature supports a coherent revision of the older review: Freeman-Sheldon syndrome is now more accurately synthesized as Freeman-Burian syndrome, a rare congenital craniofacial myopathy with characteristic facial criteria, variable limb and spinal involvement, and substantial multidisciplinary management needs.**Diagnostic precision**, clearer separation from Sheldon-Hall syndrome, and a more critical reading of inherited assumptions in the anesthetic and case-report literature still remain the key areas of research.

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