

Reply re: Periocular Anomalies in Freeman-Sheldon Syndrome

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Abstract

In Freeman-Burian syndrome (formerly, Freeman-Sheldon syndrome), correcting visual obstruction and strabismus, when present, is the operative priority and is not outweighed by anesthesia challenges; operative delay risks vision loss and developmental delay.

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To the Editor: We read with interest, “Periocular Anomalies in Freeman-Sheldon Syndrome,” published in your *Journal*.^[1] It is wonderful seeing this exquisitely rare syndrome discussed, but inaccuracies in the article could deleteriously affect patient care.

While known by many names through the years, Freeman-Sheldon syndrome is now known as Freeman-Burian syndrome (FBS) to avoid confusion with the similar-appearing Sheldon-Hall syndrome (SHS).^[2] For many years, these syndromes were thought to be a single condition, but FBS is now known to be a complex craniofacial myopathy, with SHS being a distal arthrogryposis.^[3]

In FBS, threats to vision are caused by variable replacement of normal muscle by tendinous-like constrictive fibrose tissue that causes blepharophimosis, blepharoptosis, and strabismus.^[4] In our experience, blepharoptosis is best addressed using a static sling to the frontalis, which may have extensive fibrose replacement.^[5] Secondary changes to the craniofacial skeleton are seen, but other ocular pathologies have not been observed.

As one-third of patients with a diagnosis of FBS may have been misdiagnosed, authors should demonstrate that patients fulfill the diagnostic criteria (microstomia, pursed lips, deep nasolabial folds, and H

or V-shaped chin defect and two major arthrogryposes—typically, camptodactyly with ulnar deviation and equinovarus).[6] Unfortunately, the authors neither describe their patient’s diagnostic findings nor include a facial image showing the pathognomonic findings.[1]

While fear of anesthesia and surgery is very understandable, families should be reassured that patients with FBS can be safely cared for and do not have an elevated risk for malignant hyperthermia, the hypothesis for which arose from a report of two cases.[7-8] In FBS, hyperpyrexia may occur in any physiologically stressful situation but responds to ibuprofen.[7] While the anesthesia care in FBS may be complex, correcting visual obstruction and strabismus, when present, is a priority, and delaying operative amelioration risks vision loss and developmental delay.[4,7] Challenges do not preclude safe surgeries, and a clinical practice guideline for anesthesia care in FBS is available to aid in planning.[8]

While we applaud thoughtful discussion of FBS, caution is needed. As much of the older information concerning FBS was not accurate but continues to be cited, great care must be taken when selecting articles.[6] We have reviewed eleven case reports published in the past two years which share similar problems that could have been easily prevented by searching recent literature.

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