Dear editor,

We read with great interest the article by Wróblewska-Seniuk, Jarząbek-Bielecka, and Kędzia titled, “Freeman-Sheldon syndrome - a course of the disease from birth to adulthood” [1]. The longitudinal history of a 25-year-old woman, reported as having Freeman-Sheldon syndrome [Freeman-Burian syndrome (FBS)] and undergoing labiaplasty for labia minora hypertrophy, is described [1, 2]. It is excellent to see this exquisitely rare syndrome discussed. Unfortunately, this article is somewhat unclear, and with omission of relevant recent literature, potentially harmful ideas are repeated, which this letter seeks to correct.

Without photographs or a detailed description of how the patient met the diagnostic criteria, it is not certain she had FBS. Stating the patient had FBS is insufficient, considering the false positive rate may be between 30-60% [3]. Stating the patient had FBS is insufficient, considering the false positive rate may be between 30-60% [3]. It is also unclear if the authors participated in the longitudinal care. If a retrospective records review was done, it should be stated. Medical records may be incomplete, and they lack intangible information.

In discussing clinical features of FBS, they list common features, including some from the diagnostic criteria [1, 3–5]. They do not, however, state the clinical diagnostic criteria [1, 3–5]. They place a great emphasis on distal extremity contractures, which are a non-diagnostic finding in FBS and common in many syndromic and non-syndromic entities [1, 3–6]. Not directly stating the diagnostic criteria can confuse the reader unfamiliar with FBS.

While FBS has had many classifications since its first description in 1938 [7], it appears to be a complex congenital myopathic craniofacial syndrome, as arthrogrypotic findings are not pathognomonic [3, 6]. In the syndrome, “bone anomalies” are secondary effects of the primary myopathic process of fibrose tissue replacement of normal muscle fibers (not increased muscle tone) [1, 3, 8, 9]. This fibrose tissue acts as constricting bands, the way collagen behaves in severe burns [3, 8, 9]. This is correlated with in vitro molecular myophysiology observations showing problems with the metabolic process for contraction and extreme muscle stiffness that reduces muscular work and power [10–12]. Misunderstanding of etiology in FBS has led to inappropriate treatment plans, especially surgeries, and has resulted in tragic, lifelong impairments [3, 8, 9, 13].

Cases once believed to represent an autosomal recessive or X-linked inheritance pattern are now, based on new evidence, believed to be a germline mosaicism [8]. Most inherited cases are autosomal dominant, however [3, 8]. “Sporadic” refers to a trait not having been inherited and is not associated with “causes”. Both inherited (as autosomal dominant) and non-inherited (sporadic) cases have been shown to have MYH3 mutations [5]. Ultrasound for prenatal FBS diagnosis is very limited and not possible before 20 weeks [8]. No data on life-expectancy exist [3, 8].

Anesthesia in FBS is very difficult and poses many risks [14]. While a malignant hyperthermia (MH) event is always life-threatening, the potential association of MH and FBS was based on a single report of two cases, but non-MH hyperthermia may commonly occur in FBS patients [14].

Finally, the authors confuse “esthetic” for “reconstructive” surgery, mental retardation for developmental delay caused by physical anomalies, and problems “in the eyes” for “involving the eyes” [8, 9]. Nonetheless, this article adds to the discussion of FBS, while illustrating the perils of describing a rare condition.

Abbreviations
FBS, Freeman-Burian syndrome; MH, malignant hyperthermia.

Author contributions
MIP and CRD drafted the letter together. Both authors read and approved the final manuscript.

Ethics approval and consent to participate
Not applicable.

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Conflict of interest
The authors declare no conflict of interest.

References