

LETTER TO THE EDITOR

Open Access



Comment on: anesthetic management of a pediatric patient with Freeman-Sheldon syndrome undergoing atrial septal defect closure: a case report

Mikaela I. Poling^{1*}  and Craig R. Dufresne¹

To the Editor

We read with interest a recently published case of general anesthetic management of an 8-year-old girl who was stated to have Freeman-Sheldon syndrome and underwent atrial septal defect closure [1]. While the authors are to be applauded for their excellent anesthetic management of a patient with a complex presentation, the clinical implications of inaccuracies about the syndrome contained in their report are concerning.

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, FBS and SHS were considered a single entity and part of the limb deformity group of disorders known as distal arthrogryposes (DA) [2–4]. FBS, which preferentially affects craniofacial development, is now known to be distinct from SHS, which affects the extremities and spine preferentially [2–5]. Although both are muscle disorders, FBS is considered a complex craniofacial myopathy, with SHS considered a DA [3]. While FBS shows significant variability in severity, it is a single syndrome and neither a group of conditions nor a disease.

As suggested by the literature, FBS has a 30–60% false-positive diagnosis rate [4, 5]. On closer review, it is not surprising the patient appeared to have anatomic hallmarks of SHS and a history supporting this diagnosis over FBS [1]. Facial photographs show a triangular face and a small mouth, and the history includes ease of laryngoscopically introduced endotracheal intubation [1, 4]. Patients with FBS have elongated faces and true microstomia that prevents the use of a laryngoscope [4]. Unfortunately, the authors do not clearly specify 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) [1, 4].

Patients with FBS do not have an elevated risk for malignant hyperthermia or neuroleptic malignant syndrome, the hypothesis for which arose from one report of two cases and another reporting a single patient [6–8]. In FBS, hyperpyrexia may occur in any physiologically stressful situation but responds to ibuprofen [8]. The authors do discuss several relevant aspects of anesthesia care in patients with FBS, such as avoiding respiratory depressing agents and difficult vascular access and airway management [1]. Up-to-date anesthesia care recommendations are available online through OrphanAnesthesia (orphananesthesia.eu) for FBS, SHS, and DA types 1 and 3 [8–11].

This comment refers to the article available online at <https://doi.org/10.1186/s40981-023-00633-9>.

*Correspondence:

Mikaela I. Poling
luikart2@gmail.com

¹ 8501 Arlington BLVD, Suite 420, Fairfax, VA 22031, USA

Much of the older information concerning FBS is not accurate and continues to be cited in the literature and relied upon in clinical practice. To advance the care for these very complex patients, caution must be taken when selecting articles. We have reviewed twelve case reports published since 2020 that share a similar set of problems easily prevented by searching recent literature.

Abbreviations

DA	Distal arthrogryposis
FBS	Freeman-Burian syndrome
FSS	Freeman-Sheldon syndrome
SHS	Sheldon-Hall syndrome

Acknowledgements

The authors wish to thank CM Poling, MS, for proofreading and critical comment; and M Pöcket, W Cloud, B Poling, D Nickell, and L Kesling and his team for technical assistance. The authors dedicate this work in grateful humility to The Reverend Alan Neal, a towering intellectual and man faith and action who died during the preparation of this manuscript.

Authors' contributions

MIP conceived the idea and wrote a draft. CRD critically reviewed the draft for important intellectual content. Both authors read and approved the final manuscript.

Authors' information

Both authors have extensive experience with Freeman-Burian syndrome and have published several manuscripts following a decade-long effort to clarify its definition, nomenclature, and classification and develop a multidisciplinary clinical care framework. CRD is a craniofacial surgeon, and MIP is his (unpaid) research assistant.

Funding

This work was unfunded.

Availability of data and materials

Not applicable.

Declarations

Ethics approval and consent to participate

Not applicable.

Consent for publication

Not applicable.

Competing interests

The authors declare that they have no competing interests.

Received: 14 October 2023 Revised: 14 October 2023 Accepted: 26 October 2023

Published online: 23 January 2024

References

- Takahashi K, Sakurai K, Hamaya I. Anesthetic management of a pediatric patient with Freeman-Sheldon syndrome undergoing atrial septal defect closure: a case report. *JA Clin Rep.* 2023;9(1):40. <https://doi.org/10.1186/s40981-023-00633-9>. Published 2023 Jul 13.
- Poling MI, Dufresne CR. Revisiting the many names of Freeman-Sheldon syndrome. *J Craniofac Surg.* 2018;29(8):2176–8. <https://doi.org/10.1097/SCS.0000000000004802>.
- Poling MI, Dufresne CR. Head first, not feet first: Freeman-Sheldon syndrome as primarily a craniofacial condition. *Cleft Palate-Craniofac J.* 2018;55(5):787–8. <https://doi.org/10.1177/1055665617753482>.
- Stevenson DA, Carey JC, Palumbos J, Rutherford A, Dolcourt J, Bamshad MJ. Clinical characteristics and natural history of Freeman-Sheldon syndrome. *Pediatrics.* 2006;117(3):754–62. <https://doi.org/10.1542/peds.2005-1219>.
- Poling MI, Dufresne CR, Chamberlain RL. Findings, phenotypes, diagnostic accuracy, and treatment in Freeman-Burian syndrome. *J Craniofac Surg.* 2020;31:1063–9. <https://doi.org/10.1097/SCS.0000000000006299>.
- Jones R, Dolcourt JL. Muscle rigidity following halothane anesthesia in two patients with Freeman-Sheldon syndrome. *Anesthesiology.* 1992;77(3):599–600. <https://doi.org/10.1097/0000542-199209000-00031>.
- Stein MH, Sorscher M, Caroff SN. Neuroleptic malignant syndrome induced by metoclopramide in an infant with Freeman-Sheldon syndrome. *Anesth Analg.* 2006;103(3):786–7. <https://doi.org/10.1213/01.ANE.0000227135.46049.B7>.
- Poling MI, Dufresne CR. Freeman-Burian syndrome. *Anästh Intensivmed.* 2019;60(1):S8–17. <https://doi.org/10.19224/ai2019.S008>.
- Poling MI, Dufresne CR. Anaesthesia recommendations for Sheldon-Hall syndrome. *OrphanAnesthesia.* 20 Feb 2019. Available at: https://www.orphananesthesia.eu/en/rare-diseases/published-guidelines/cat_view/61-rare-diseases/60-published-guidelines/237-sheldon-hall-syndrome.html.
- Poling MI, Dufresne CR. Anaesthesia recommendations for distal arthrogryposis type 1. *OrphanAnesthesia.* 24 Feb 2019. Available at: <https://www.orphananesthesia.eu/en/rare-diseases/published-guidelines/distal-arthrogryposis-type-1.html>.
- Poling MI, Dufresne CR. Distal arthrogryposis type 3. *Anästh Intensivmed.* 2019;60(13):S536–43. <https://doi.org/10.19224/ai2019.s536>.

Publisher's Note

Springer Nature remains neutral with regard to jurisdictional claims in published maps and institutional affiliations.

Submit your manuscript to a SpringerOpen® journal and benefit from:

- Convenient online submission
- Rigorous peer review
- Open access: articles freely available online
- High visibility within the field
- Retaining the copyright to your article

Submit your next manuscript at ► [springeropen.com](https://www.springeropen.com)