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DC PLASTIC SURGEON TACKLES FAKE NEWS ABOUT RARE DISEASE, FREEMAN-BURIAN SYNDROME, IN ONLINE ARTICLES

Fairfax—Continuing his efforts to confront common myths about Freeman-Burian syndrome (FBS) (previously, Freeman-Sheldon syndrome) and improve care for patients with this very rare condition, Dr Craig R Dufresne (<https://cdufresnemd.com/plastic-surgery-office/dr-dufresne/>) published two preprint articles on *Authorea*, fact-checking yet more previously published medical journal articles. Dufresne, a world-renowned rare disease specialist and DC area plastic surgeon, has decades of experience caring for patients and writing about FBS. FBS, an exceptionally rare and difficult to treat birth defect, is primarily a condition of facial and skull muscles that frequently involves muscles in the arms, legs, and elsewhere.

On December 30, 2024, “Patients with Freeman-Burian Syndrome can Experience Unrelated Ailments,” (<https://doi.org/10.22541/au.173557839.92597332/v1>) was published in response to a December 19, 2024 article (<https://doi.org/10.7759/cureus.76037>) discussing trabeculotomy using the Kahook Dual Blade in a patient said to have FBS. In his response, Dufresne cautioned that the false-positive diagnosis rate for FBS was high and a correct diagnosis should always be ensured. Dufresne also reminded readers that just because a patient has a major diagnosis, such as FBS, that does not preclude the possibility of unrelated problems.

Previously, on July 1, 2022, “Reply re: Periocular Anomalies in Freeman-Sheldon Syndrome,” (<https://doi.org/10.22541/au.165668902.20562659/v1>) was published in response to a June 2022 article (<https://doi.org/10.1097/IOP.0000000000002207>) describing eyelid problems and anesthesia concerns for patients with FBS. Dufresne’s preprint provides an update to anesthesia best-practices and cautions that delaying eyelid surgery for infants and young children could result in vision loss, if light entering the eye is obstructed.

Preprints are freely available articles but have not been critically evaluated by other doctors and editors. Preprints typically contain information authors wish to make public quickly, without waiting for a medical journal to finish processing their article. It’s Dufresne’s hope wider dissemination of accurate and up-to-date information will improve patient care.

For more information and to arrange interviews, contact Ms Mikaela Poling, *Research Fellow*, research@duplastics.com or (304) 460-9038.

Craig R Dufresne, MD, PC, in Fairfax, Virginia, is a premier private solo practice providing aesthetic and reconstructive surgery care to adults and children from across the globe. Research supports the mission to provide safe, exceptional, innovative, and compassionate care that enhances overall well-being and health.

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