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FACT-CHECKING ARTICLE IN EUROPEAN MEDICAL JOURNAL CORRECTS THE RECORD ON RARE CRANIOFACIAL DISORDER

FAIRFAX—The October issue of the *Clinical and Experimental Obstetrics and Gynecology*, published online Friday, features a letter (https://ceog.imrpress.com/EN/10.31083/j.ceog4805160) by world-renowned rare disease specialist and DC area plastic surgeon, Dr Craig R Dufresne, addressing common misconceptions concerning Freeman-Burian syndrome (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. Dufresne's letter is in response to a previously published article (https://ceog.imrpress.com/EN/10.31083/j.ceog.2020.06.5430) in the *Journal*, whose authors he felt had included inaccuracies that were potentially dangerous to patients.

In his letter, Dufresne reviewed diagnostic criteria of the syndrome, which, he explained, centered on the presence of certain facial features, not hand or foot deformities, as the other authors suggested. He also cautioned, "the false positive rate [of FBS] may be between 30-60%." Dufresne particularly emphasized the importance of understanding the condition arises from problems in the muscles, not, "'bone anomalies'[, which] are secondary effects of the primary myopathic process of fibrose tissue replacement of normal muscle fibers (not increased muscle tone)." Dufresne continued, saying: "[Failure to understand FBS as a muscle disorder] has led to inappropriate treatment plans, especially surgeries, and has resulted in tragic, lifelong impairments."

Finally, Dufresne clarified that FBS was believed to only be passed on from parent to child in an autosomal dominant manner, only *MYH3* gene mutations have been found to cause FBS, prenatal ultrasound for FBS diagnosis was unreliable, anesthesia for patients with FBS was very difficult and posed many risks, and the rare life-threatening anesthesia reaction of malignant hyperthermia was not believed to be associated with FBS.

It's Dufresne's hope wider dissemination of accurate and up-to-date information will encourage a total paradigm shift and herald a new era of vastly improved patient care and greater understanding of FBS by patients, families, and others. For Dufresne, writing about rare conditions is all about educating scientists, physicians, care teams, family members, and patients to improve patients' chances for a healthy, normal, and productive life. An ever-humble gentleman, publishing medical articles is his way to help many more patients than he ever could directly.

For more information and to arrange interviews with Dufresne and a patient with Freeman-Burian syndrome, contact Ms Mikaela Poling, *Research Assistant*, research@duplastics.com or (304) 460-9038; or Mr Christopher Dufresne, *Office Manager*, info@cdufresnemd.com or (703) 207-3065.

Craig R Dufresne, MD, PC, with offices in Fairfax, Virginia and Chevy Chase, Maryland, 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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