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PRESS RELEASE

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NIH GENETIC ONLINE ENCYCLOPEDIA ARTICLE IMPROVED WITH RESEARCH OF RARE DISEASE EXPERT AND DC PLASTIC SURGEON

FAIRFAX—On October 28th, National Institutes of Health's Genetic Home Reference, now part of MedlinePlus, published their revised their encyclopedic entry on Freeman-Sheldon syndrome (https://medlineplus.gov/genetics/condition/freeman-sheldon-syndrome/) based on the published research of world-renowned DC area rare disease specialist and plastic surgeon, Dr Craig R Dufresne, MD, FACS, FICFS. The move follows earlier encyclopedic revisions made by the NIH's Genetic and Rare Diseases Information Center (GARD), Genetic Alliance, and National Organization for Rare Disorders. Freeman-Sheldon syndrome, more recently Freeman-Burian syndrome (FBS) is primarily a condition of facial and skull muscles that frequently involves muscles in the arms, legs, and elsewhere. Incorporating information from Dufresne's research has made the entry more complete and current, though Dufresne remains concerned about the accuracy of some of the information contained in the NIH article.

First, Dufresne says the revised encyclopedic entry did not clearly state the diagnostic criteria, despite including an enumeration of the required features. Second, the article does not specify the limited circumstances under which mental retardation can occur in FBS, namely following severe neonatal hypoxia. Dufresne cautions that the statement that the article makes that, "Intelligence is unaffected in most people…" leaves families feeling that their child or loved one could have some type of intellectual disability as part of FBS. Dufresne explains, "It doesn't work like that; patients with FBS are at greater risk of hypoxia in the neonatal period, which can cause intellectual impairment if it's prolonged." He continues, "Intellectual disabilities, in and of themselves, are not associated with FBS, however." Third, the reference list continues to include an article about distal arthrogryposes, again adding confusion about FBS's classification, according to Dufresne. Dufresne asserts FBS is a craniofacial syndrome, which is clearly echoed in the revised article. The article remains unclear in other areas, as well.

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, speaking and 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, presenting and publishing medical research 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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