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NIH FLAGSHIP ONLINE RESOURCE IMPROVED WITH RESEARCH OF RARE DISEASE EXPERT AND DC PLASTIC SURGEON

FAIRFAX—Recently, the National Institutes of Health flagship information resource for rare diseases, Genetic and Rare Diseases Information Center (GARD), revised their entry on 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. 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.

"FBS is a craniofacial syndrome that involves the muscles," Dufresne says, "but their article still talks about it as a bone and joint condition, which isn't accurate." Dufresne continued: "In FBS, white fibrous tissue replaces normal muscle tissues in some areas. It behaves like scar tissue, forming constricting bands." Dufresne says it is these "constricting bands" that indirectly cause the joint problems and deformed-appearing ones. He explains that bones take their shape from forces exerted on them by muscles, so when those forces are abnormal or non-existent, the bone shape differs from "the norm".

Dufresne is a clinical researcher, not a basic science researcher, but he points to basic science work done by others that support what he sees during surgery and on microscope slides of his patients' muscles. "The data point toward disturbances in the metabolic process for contraction and toward extreme muscle stiffness that reduces muscular work and power," he says.

For support for FBS's effects being skewed toward the face and skull, Dufresne references his recent study of all published cases—the largest to-date—that appeared in the *Journal of Craniofacial Surgery* and was cited in the NIH article. Though a study of previously published data, he says his findings were supported by smaller studies of newly collected data done earlier by a separate group.

There are other concerns, too. Perhaps, the most worrying for families and patients' is the NIH article's suggestion FBS may be a progressive condition. Dufresne says there is no evidence of this.

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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