

## **University of Washington**

*"To Operate or Not to Operate?" Stigma and Quality of Life Outcomes in Craniofacial Fibrous Dysplasia*

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Fibrous dysplasia is a rare mosaic disease of bone marrow stromal cells, caused by post-zygotic somatic activating mutation in the gene *GNAS*. Normal bone and marrow are replaced by fibro-osseous tissue, which may lead to fractures, deformity and pain. The population with craniofacial fibrous dysplasia (CFD), including lesions in the face, jaws, skull or skull base, is the largest subgroup. Expansile bony lesions may lead to facial asymmetry and distortion of features, sinus or dental problems, or rarely, impairment of vision or hearing.

Few clinicians outside tertiary care centers have experience with CFD, and most literature is in the form of case reports (CR). CR indicate that craniofacial surgery should be performed to treat functional problems or for cosmetic purposes, to mitigate patient's experience of social stigma and improve self-esteem. Surgical interventions include bone biopsy, re-contouring procedures, and resection +/- reconstruction. The National Institutes of Health natural history study has examined cases of CFD surgery, noting that regrowth after surgery is common, with subjects requiring multiple operations, leading to pain and other morbidity. This suggests that observation is also a viable treatment option, with annual monitoring of lesions to maintain function.

There are no comprehensive studies regarding the indications for operative treatment versus medical management and which treatment choice is more effective in the long-term. Furthermore, reports of quality of life (QoL) of individuals with CFD are lacking, and the impact of surgery on QoL is not established. Previous NIH work determined that life satisfaction did not correlate with quantitative FD disease burden. Some qualitative research also suggests that the objective amount of disfigurement does not drive decisions to have surgery. Thus, we hypothesize that CFD subject dissatisfaction and QoL are not correlated with the amount of disease.

In CFD, stigma is a component of the disease experience – because distortion of the face and skull are jarring to others and because others may associate distortion with intellectual deficits. Patients with CFD negotiate negative reactions to their appearance and their own awareness and feelings about deviation from “normal.” Perceived stigma from others and self-stigma are associated with negative mental health outcomes, including depression, low self-esteem, and self-isolation. Surgically removing the causes of stigma could be viewed as a treatment for these psychological effects. However, previous research into other craniofacial diseases report that surgery may not result in patient satisfaction and resolution of psychological discomfort.

Our study will answer the PICO question: Among patients with CFD, do those treated operatively, when compared to those managed nonoperatively, have improved QoL and less stigma? Our overall goal is to assist CFD patients with informed-decision making and to provide better standards of care regarding surgery.

We will collect CFD patient-reported data through the Fibrous Dysplasia Foundation Patient Registry (FDFPR). This online patient portal currently uses validated measures to gather CFD respondent information regarding disease diagnosis, demographics, self-reported motivations for surgery, types of operations and time-frame, use of medications, pain, function and aspects of QoL. It enables

comparison of the surgical and observational population. We will examine satisfaction and QoL measures, comparing subjects who have chosen observation vs those who have chosen craniofacial surgery. Confounding factors such as use of pain medication, types of surgical practices, number of surgeries, and counseling will be controlled. For this cohort study, our anticipated sample size is 100.