

Facioscapulohumeral (FSH) dystrophy is a common muscular dystrophy in which there is progressive weakness of the face, upper arms and shoulder regions as well as the legs. The symptoms of FSH dystrophy may appear during childhood with severe facial and limb weakness or develop slowly and gradually in adulthood with difficulty such as eye closure, lifting or tripping. The disease is caused by degeneration of muscle due to a specific chromosomal deletion. This deletion is inherited from one generation to the next. FSH dystrophy is the third most common dystrophy with an estimated prevalence of 1 in 20,000. In spite of this high prevalence, research and treatment options are at a suboptimal level. We are collaborating with neurologists and therapist who focus in the care of this disease. We have focused on facial manifestation of FSH dystrophy and made several observations that have lead to treatment options for our patients.

The muscles of facial animation are small skeletal muscles that mostly originate and insert on the soft tissues of the face. Facial expression is an important part of human communication allowing us to reflect emotions and project non-verbal cues. The facial muscles also support the facial skin and fat giving it shape and form. Blinking, whistling, blowing out candles, drinking from a straw are simple day to day actions that depend of the fine action of the facial muscles. These functions are severely affected when the facial muscle become progressively weak as seen in FSH dystrophy. FSH dystrophy patient often complain of changes in their speech. They are unable to generate the intraoral pressure to pronounce plosive sounds such as "P" "B". As the facial muscle become weaker, midfacial fat descends the corner of the mouth turns downwards giving a falsely sad appearance.

Muscle tone and movement are two attributes to consider when exploring treatment option for facial weakness related to FSH dystrophy. Muscles in the body are maintained at a slightly contracted state which is called the muscle tone. The always activated state of partial contraction keeps the muscles firm, offers some resistance and maintains form and posture. Facial muscle tone keeps the face from drooping, eyelids from sagging keeps the lips together and offers some cheek resistance when preparing to blow a candle. When the facial muscles become dystrophic, they lose tone and will become flaccid. To restore tone, weak facial muscles should be regenerated or replaced. Regenerative medicine and tissue engineering research is presently a major focus of many researchers and may in the future offer options for reconstituting dystrophic muscle. Muscle movement is needed for facial expression and sphincter function around the eyes and mouth. Although the facial nerves are intact, dystrophic facial muscles do not respond well and animate the face in a very limited manner. Replacing a dystrophic muscle is technically feasible using microsurgical techniques. Microsurgery now allows us to borrow a section of muscle from one part of the body and transplant it to another part to generate a desired effect. The main limitation in pursuing muscle replacement surgery as treatment for facial FSH dystrophy is the progressive nature of the disease and the number of potential donor muscles that can be affected. In other words, why replace a weak muscle with another muscle that may also become weak. One solution is to determine potential donor muscles that are not commonly affected in FSH dystrophy. In the head, the temporalis muscle appears to be spared in FSH dystrophy. We have successfully transferred the temporalis muscle with techniques uniquely tailored to FSH dystrophy patients. We are investigating other potential donor muscle. The ideal donor muscle should be available for harvest without any significant functional loss and should be spared from progressive dystrophy. Such a muscle can be transferred to the face and reinnervated with the existing facial nerve. Facial tone can be improved through other static means. The basis of these static procedures is to introduce some degree of form, shape, stiffness and resistance to the flaccid areas of weakened face. Collagen matrix grafts strategically placed in the lower eyelid can lift the eyelid vertically up and allow better eyelid closure. Fat grafting to the buccal space over the flaccid buccinators muscle also introduce some resistance to the cheek which helps with plosive sounds. Targeted lip augmentation together with muscle placcation techniques can be used to correct lip position, shape and closure. The current options for reanimating the face in FSH dystrophy are promising but should be approached carefully under the direction of a multidisciplinary team that specializes in this disease.