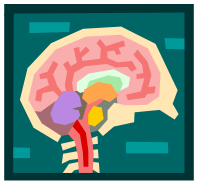

Disability Awareness Begins With You: Freeman-Sheldon Syndrome

Freeman-Sheldon Syndrome (also known as "Whistling Face Syndrome") is a rare genetic condition which characteristically includes a small "whistling" mouth, a flat mask-like face, club feet, joint contractures usually involving the fingers and hands, and under-development of the cartilage of the nose. Intelligence is often normal. Most of the features of this syndrome are due to muscle weakness.

Freeman-Sheldon Syndrome is most recognizable by its facial features. The hallmark of the syndrome is the small mouth.

Mouth: The mouth is small, with tight, pursed lips that look the person is attempting to whistle. This can make feeding more difficult, and can pose a choking hazard if the obstruction cannot be easily removed. The small mouth can be enlarged through surgery.

Eyes: The eyes can appear a bit sunken, and the eyelids may be somewhat drooped. This can impair vision, but can be corrected through surgery. In addition, someone with FSS may appear to be cross-eyed ("Strabismus").

Long Philtrum: The distance between the nose and mouth is extended.

Chin: "H" or "Y" shaped dimpling on the chin.

High palate: The high palate can affect speech, resulting in a more nasal quality.

In addition, people with Freeman-Sheldon Syndrome have orthopedic problems.

Clubbed Feet: Feet can be turned outward or inward, hampering the ability to walk. This can be corrected through casting or surgery. In extreme cases, amputation may be necessary .

Scoliosis: A curved spine, either bowed or in an "S" shape, can compress heart and lungs if left untreated. The progression of the curve can be slowed through bracing, but surgery is required in most cases.

"Wind-vaned" or Clinched Hands: Hands can point outward or be clinched in fists. This impairs hand function, but can be corrected with splints, occupational therapy, and/or surgery in most cases.

Malignant Hyperthermia: A severe reaction to inhaled surgical anesthesia, where the muscles seize up and body temperature spikes. Malignant Hyperthermia has occurred in some individuals with FSS.

Freeman-Sheldon syndrome (FSS) is a multifaceted condition involving skeletal muscle tissue and tissue attached to muscle or affected muscle tension at all locations throughout the body. Because the syndrome varies, standards for care must be sufficiently broad to encompass each individual's specific malformations and disabilities.

- Surgical procedures to correct feeding dysfunction (such as gastrostomy) and breathing difficulties (tracheotomy).
- Speech therapy and/or dysphagia team intervention to correct pharyngeal dysfunction and/or feeding difficulties.
- Speech therapy for delayed and/or nasal speech.
- Communications boards for those unable to speak.
- Dental and oral prostheses to correct microstomia and/or hypoplastic mandible.
- Orthodontia.
- Plastic surgery to correct malformations of the face and hands.
- Orthopedic surgery to correct clubfeet, joint contractures and scoliosis.
- Braces and wheelchairs to aid mobility.
- Physical therapy.
- Occupational therapy.
- An ophthalmologist to correct strabismus and/or ptosis (eyelid droop).
- Auditory testing with possible need for hearing aids
- A neurologist to evaluate and manage any developmental delay which may occur.
- Genetic counseling.
- Psychological counseling to better cope with facial malformation and orthopedic disabilities.
- Early intervention programs.
- Special education programs as needed

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