Fibrodysplasia ossificans progressiva (FOP)

Fibrodysplasia ossificans progressiva (FOP) is a rare, genetic condition that is disabling to the congenital skeletal due to ossification. This disease causes muscles and soft connective tissue to turn into bone, called heterotopic ossification (HO). There is no cure for this disease. There are two clinical features of Fibrodysplasia ossificans: big toe malformations and specific spatial patterns of progressive HO. Children with this disease appear normal at birth; however, in the first 10 years of life they began to develop painful, high inflammation of the soft tissue, which causes swelling. This swelling causes the soft connective tissue to transform or become encased in bone. Soft connective tissue includes the following:

- **Aponeuroses**: “A sheetlike fibrous membrane, resembling a flattened tendon, that serves as a fascia to bind muscles together or as a means of connecting muscle to bone.” (Quinn, 2009)
- **Fascia**: “The superficial fascia is a soft connective tissue located just below the skin. It wraps and connects the muscles, bones, nerves and blood vessels of the body. Together, muscle and fascia make up what is called the myofascia system.” (Professional Health Systems, Inc., 2013)
- **Ligaments**: “Ligaments connect bone to bone.” (BME/ME 456 Biomechanics, n.d.)
- **Tendons**: “The tendons are tough, whitish cords, varying in length and thickness, and devoid of elasticity. They consist almost entirely of white- fibrous tissue.”
- **Skeletal muscles**: “Skeletal muscle is striated muscle tissue that is attached to bones. It is composed of fibers that look like a mixture of dark and light bands bundled together that run along the bone.” (Conjecture Corporation, 2013)

According to Professional Health Systems, Inc. (2013), “FOP involvement is typically seen first in the dorsal, axial, cranial and proximal regions of the body and later in the ventral, appendicular, caudal and distal regions. Several skeletal muscles including the diaphragm, tongue and extra-ocular muscles are enigmatically spared from FOP. Cardiac muscle and smooth muscle are not involved in the FOP process.”

The cause of this disease is a genetic mutation in the gene that “provides the body with instructions on how to produce the protein that is part of the protein family called bone morphogenetic protein. This protein helps regulate the growth and development of muscles and bones and helps control the gradual replacement of cartilage by bone that occurs when a person grown in to an adult” (Wellness.com, 2013).

FOH can occur randomly or be passed genetically. Typically, nearly all cases are random occurrences; however, when inherited genetically it happens through an autosomal dominant trait. This means that only one parent needs to have the gene in order for the child to inherit this disorder. Generally, this is rare though because the majority of the people with this disease are unable to have children.

### Symptoms (Wellness.com, 2013)

- Malformed toes that turn outward
- Painful, tumor like swellings in the neck, shoulders and back (eventually becomes ossification)
- Painful flare-ups when new bones develop
- Stiff joints
- Bedridden by age 30
- Death occurs before age 40
- Nearly 50% experience conductive hearing loss due to bone ossification of the ossicular chain, some however experience sensory neural hearing loss.

### School Environment:

Students with FOP typically have an aid to help protect them from having trauma to their bodies, like bumps or falls since trauma can cause bone growth. The aid also assists with getting books, turning pages, and maneuver books so that the student can see them.
Resources:

Web MD (2012) list the following resources:

**International FOP Association**
P.O. Box 196217
Winter Springs, FL 32719-6217
Tel: (407)365-4194
Fax: (407)365-3213
Email: together@ifopa.org
Internet: http://www.ifopa.org

**March of Dimes Birth Defects Foundation**
1275 Mamaroneck Avenue
White Plains, NY 10605
Tel: (914)997-4488
Fax: (914)997-4763
Email: Askus@marchofdimes.com
Internet: http://www.marchofdimes.com

**NIH/National Institute of Arthritis and Musculoskeletal and Skin Diseases**
Information Clearinghouse
One AMS Circle
Bethesda, MD 20892-3675
USA
Tel: (301)495-4484
Fax: (301)718-6366
Tel: (877)226-4267
TDD: (301)565-2966
Email: NIAMSIinfo@mail.nih.gov
Internet: http://www.niams.nih.gov/

**MUMS National Parent-to-Parent Network**
150 Custer Court
Green Bay, WI 54301-1243
USA
Tel: (920)336-5333
Fax: (920)339-0995
Tel: (877)336-5333
Email: mums@netnet.net
Internet: http://www.netnet.net/mums/

**Genetic and Rare Diseases (GARD) Information Center**
PO Box 8126
Gaithersburg, MD 20898-8126
Tel: (301)251-4925
Fax: (301)251-4911
Tel: (888)205-2311
TDD: (888)205-3223
Internet: http://rarediseases.info.nih.gov/GARD/AboutGARD.aspx

Citations:


