The genetic disorder of fibrodysplasia essay sample

The genetic disorder of fibrodysplasia ossificans progressiva has only been recorded about 2500 times since the 1800’s and believed to occur in approximately 1 in 2 million people worldwide (Molecular Consequences of the ACVR1R206H Mutation of Fibrodysplasia Ossificans Progressiva*). Fibrodysplasia ossificans progressiva or FOP is a disorder that makes muscle and connective tissue gradually get turned into bone which then in turn inhibits motion. Individuals that are born with FOP are generally born with malformed big toes, short thumbs and other skeletal abnormalities.

These anomalies in characteristics help distinguish FOP from other bone and muscle problems. The extra- skeletal formation that is caused by a mutations in the ACVR1 gene which instructs a member of the protein family, bone morphogenetic protein or BMP type I receptors (Mutant Activin-Like Kinase 2 in Fibrodysplasia Ossificans Progressiva are Activated via T203 by BMP Type II Receptors). This gene controls the growth and development and bones and muscles which have been found can be turned off or on that causes gradual replacement in cartilage during maturation or in FOP causes bone growth flair ups.

So FOP is overall linked to a recurrent “ mutation in activin receptor IA/activin-like kinase 2 (ACVR1/ALK2), a bone morphogenetic protein (BMP) type I receptor, is the cause of all sporadic and familial cases of classic FOP” (The Natural History of Flare-Ups in Fibrodysplasia Ossificans Progressiva (FOP): A Comprehensive Global Assessment). Most of the time FOP is caused by new mutations in those genes but this condition can also be inherited in an autosomal dominant pattern (50% chance of a child developing the disease if 1 parent is positive).

This disorder is seen all over the world and not limited to or seen more in any race or ethnicity and is most likely due to poor reproduction fitness and mutations in the ACVR1 gene. Now with patients themselves with this disorder they tend to see “ flair ups” or bone growth in the soft tissues beginning in the back, head, neck, and shoulders, and progressively then effect chest, hips, knees, and lower extremities.

Oddly, FOP spares some tissue, “ including the diaphragm, tongue, extra-ocular muscle, cardiac muscle, and smooth muscle” (http://www. medicalbag. com/profile-in-rare-diseases/fibrodysplasia-ossificans-progressiva/article/472275/). The heterotopic ossification can be triggered by trauma to muscle or connective tissue, such as, bruises, or falls, surgery, intramuscular vaccination and etcetera.

This makes for testing or any kind of treatment difficult because taking samples, having surgery, and biopsies result in swelling, inflammation, and flare-ups that make things worse. With individuals with this disorder they eventually end up motionless because all their connective tissue, ligaments, tendons, fasciae, and aponeuroses turn into bone.

Through this process usually individuals are in a wheelchair by mid-20’s and if lucky enough they can chose what position they want to be molded into (standing or sitting). The median age of death for individual living with FOP is 40 and is typically caused by TIS complications (cardiorespiratory failure), restriction of breathing, and jaw locking (not being able to eat). This rare disorder causes for individuals to eventually be entombed in their own

bodies in a certain position and no treatment to help them besides glucocorticoids that may help reduce inflammation.