INTRODUCTION

Fibrodysplasia ossificans progressive (FOP) is a rare and crippling disease. It is characterized as a condition of congenital skeletal malformation and progressive heterotopic ossification of connective tissues. This disorder was originally described by Patin in 1648 as “the woman that turned into wood”. FOP causes a progressive immobility as skeletal muscle and soft connective tissue transform into a second skeleton of heterotopic bone.

FOP is defined by two clinical features: malformation of the great toes and heterotopic ossification that is progressive and occurs in a specific spatial pattern. No heterotopic ossification is present at birth; malformation of the great toes is the only abnormality present. Typically during the first decade of life, children develop highly inflammatory, painful soft tissue swellings. These events, described as flare-ups, transform soft connective tissue into an armor-like encasement of bone.

Flare-ups, leading to progressive heterotopic ossification and progressive permanent immobility, can be triggered by minor trauma such as intramuscular injections, muscle fatigue and blunt trauma. Attempted surgical removal commonly leads to explosive and painful new bone growth.

Manifestations in the head and neck include ankylosis of the temporomandibular joint and conductive hearing loss from middle ear ossification. Patients with FOP develop thoracic insufficiency syndrome. Thoracic insufficiency syndrome leads to pneumonia and right sided heart failure.

There is no cure for FOP and medical intervention is supportive.

METHODS AND MATERIALS

The patient’s chart was reviewed in detail. An extensive literature review was performed.

RESULTS

A 2 year 7 month old boy presented with a tender, firm, left posterior neck mass. The mass came on acutely and had been recognized two days earlier by the patient’s mother. At time of presentation, the patient’s neck range of motion was limited, but his mother stated that he had always had a stiff neck.

Imaging demonstrated a non-rim enhancing fluid-like process infiltrating the posterior neck musculature, extending to the occiput. It was diagnosed clinically as an infected lymphangioma and treated with steroids and antibiotics.

Over the next two weeks he failed to show any significant improvement and the mass had now spread down to the neck and involved his left shoulder blade. Due to the concern of the rapid spread of the lesion and its clinical firmness, he underwent biopsy of the lesion. At the time of biopsy the lesion pale, firm, avascular and infiltrating the soft tissues. Final pathologic analysis demonstrated a low grade fibromyxoid and adipose tissue favoring lipofibromatosis. Diagnosis was lipofibromatosis.

The lesion continued to spread down his back, creeping around his rib cage and extending down to his hips. Further investigation revealed he had bilateral, short malformed toes with a valgus deformity. A rheumatology consultation was obtained and a diagnosis of fibrodysplasia ossificans progressiva was made based on the pathologic findings, great toe deformities and stiff joints. Genetic testing confirmed the diagnosis and the patient was started on supportive therapy.

DISCUSSION

Heterotopic ossification progresses in a reliable anatomic and temporal pattern. It follows the pattern of embryonic skeletal formation. Proximal, cranial, axial and dorsal regions of the body are typically involved first by FOP, followed by peripheral and distal spread. Cardiac muscle, smooth muscle, the diaphragm, tongue and extracranial muscles are universally spared.

Outcomes for patients include; confinement to a wheelchair by their third decade, lifelong assistance for activities of daily living, kyphosis of the jaw, and pneumonia or right sided heart failure from rigid fixation of the chest wall. Median survival is 45 and death typically occurs from complications of thoracic insufficiency syndrome.

Misdiagnosis is common, given the rarity of the disease, and failure of clinicians to associate the soft tissue swellings with malformation of the great toes. Diagnosis is typically clinical. Common misdiagnoses include; aggressive juvenile fibromatosis, lymphedema, or soft tissue sarcomas. Children frequently undergo biopsy in an effort to diagnose FOP which exacerbates the progression of the disease.

There is no cure for FOP and medical intervention is supportive. Generally surgical therapy is avoided as trauma to soft tissue exacerbates heterotopic ossification. Glucocorticoids are used during acute flare-ups, but there is no proven efficacy with any therapy in altering the natural history of the disease.

CONCLUSIONS

FOP is a clinical diagnosis with a pairing of progressive heterotopic ossification of connective tissue and malformation of the great toes.

Clinical diagnosis is imperative to avoid possibly harmful and disease exacerbating biopsy.

Treatment is supportive and prognosis for these children is grim.

REFERENCES