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A REVIEW ON STONEMAN SYNDROME

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Abstract:

Stoneman syndrome is a rare connective tissue disorder with autosomal dominant inheritance. It is caused by abnormal ectopic ossification of the tendons and ligaments of the body. The cases of stoneman syndrome are 1 in 2 million worldwide. GTR is conducted by a group of medical professionals that majorly deals with research and analysis of such rare conditions and it includes a questionnaire for both patient and the medical team. Treatment for Fibrodysplasia ossificans progressiva do not have an effective cure till now, and since the removal of the bone will only lead to the growth of new and more painful bone formation, medical professionals have come up with a couple of medications that slow down the growth and intensity of ossification, they are : Corticosteroids like Prednisone to reduce pain and sudden swelling seen in the early stages. It is recommended to be within 24hrs of the first outburst, before NSAIDS are to be given between sudden swelling Drugs that relax muscles .Mast cell inhibitors .Aminobisphosphonates .Devices such as braces or special shoes to help with walking. Occupational therapy. Those things are to be monitored that can increase the ossification and physical discomfort. Treatments and medical diagnoses associated with bone such as biopsy, Intramuscular injections (immunizations) or injections, and jaw stretching during dental procedures must be avoided, as it can damage the skeletal system which can speed up the process of ossification in the affected area. FOP is dangerous for pregnant women, childbirth must be avoided due to two reasons, firstly the genetic malfunction can be transferred to the baby which will make the child's survival difficult. Secondly, the increasing fetus in your womb can increase the risk of sudden outbursts which can be life-threatening for both baby and the mother.

INTRODUCTION

Stoneman syndrome, also called Fibrodysplasia ossificans progressiva (FOP) or Munchmeyer disease, is a rare connective tissue disorder with autosomal dominant inheritance. [1] In this disease, the condition can be characterized by abnormal ectopic ossification of the tendons, ligaments, skeletal muscles and other soft tissues of the body. [1] Smooth muscles are not involved in this disorder. Frequency of these conditions is one in two million people. [1] The disease has imaging and clinical features including bilateral hallux valgus deformity, interphalangeal big toe, heterotopic muscle and connective tissue ossification, short and wide femoral neck, pseudo exostoses, short metatarsals/metatarsals, facet joint C2-C7, large posterior elements and narrow and long vertebral bodies. [2]

Ectopic bony growths have a specific pattern of involvement, occurring in a craniocaudal, proximal distal, and dorsoventral manner. The disease is progressive and complicated by restriction of movement in corresponding areas, respiratory failure and lung infections. The common cause of death in this condition is due to cardiac and respiratory failure, which results from severe restriction of the movements of the chest wall. [2] Since the disorder has characteristic findings, it can be easily diagnosed with simple radiographs alone. The primary diagnosis of this disorder is important to avoid the necessary invasive investigations such as biopsies, as even mild trauma and intramuscular injections can exacerbate the development of a disease condition with inflammation. [3] Hence, for radiologists the knowledge of this disease condition is important to avoid invasive investigations.

DEFINATION

Stoneman syndrome or Fibrodysplasia ossificans progressiva (FOP) or Munchmeyer disease is an extremely rare genetic disorder which is characterized by ectopic ossification of the skeletal and connective tissues leading to progressive fusion of axial and appendicular skeleton. [4]

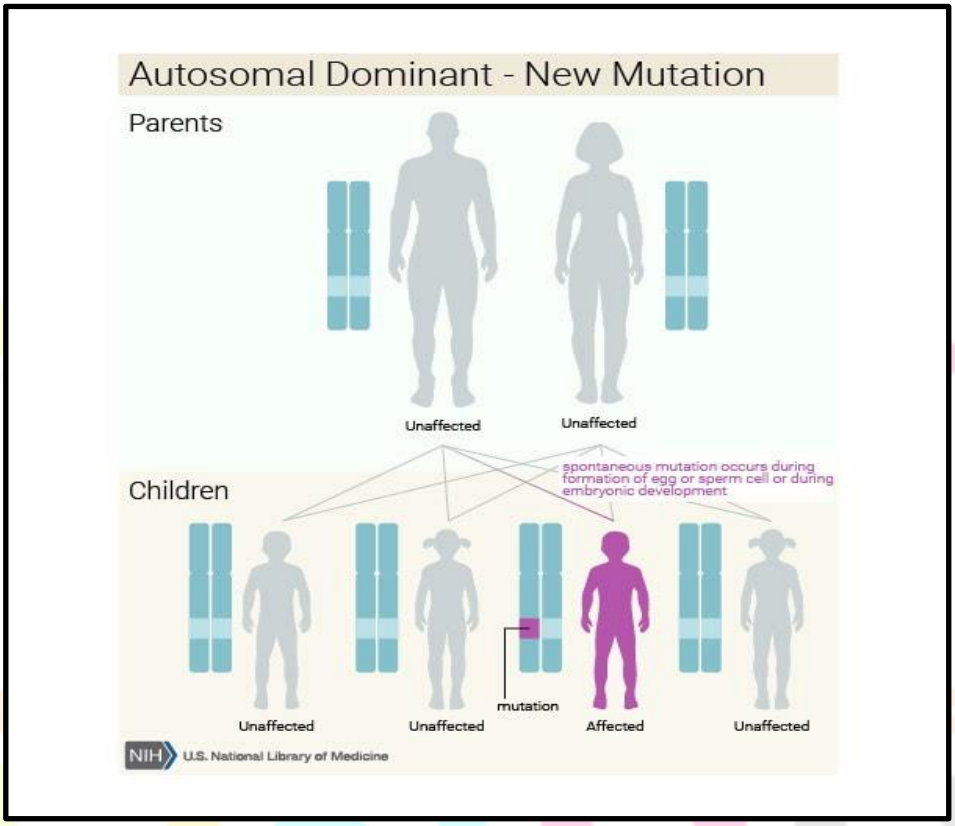
CAUSE

The ACVR1 gene is responsible for the mapping of bone morphogenetic protein (BMP) type l receptors. ACVR1 gene can be found in many tissues of the body including cartilage and skeletal muscles. BMP is a member of the protein family which maintains the right balance of morphogenesis, cellular lineage commitment, proliferation, differentiation, and apoptosis of various types of cells throughout the body.

ACVR1 gene helps in the development of muscles and bones including ossification. Ossification aids gradual replacement of cartilage by bone normal In the skeleton maturation healthy individual.

Disruption in mechanisms which control the activities of receptors can be caused due to mutations in the ACVR1 gene. The Abnormal activation of the receptor leads to increase in required ossification of BMP leads to Fibrodysplasia ossificans progressiva.
The cases of Fibrodysplasia ossificans progressiva (FOP) i.e. stoneman syndrome is very low, probably 1 in 2 million worldwide. Stoneman Syndrome's illness runs in autosomal dominant patterns, which proceed to malfunction due to presence of altered gene copy in each cell. [6]

Transfer of stoneman syndrome from parents to offspring [5]



SYMPTOMS

Stoneman syndrome is a genetic disorder so it is both easy and difficult to identify. It is a rare genetic disorder and also not known by the public so anyone can get confused with other muscular disorders. If a person or blood is related to a person who is affected by this rare genetic disorder, then these disorders can be transferred to their off-springs.

We can see changes in toes and thumbs by close examination in their newborn child. Infants with this syndrome are born with abnormal big toes and in some cases abnormal thumbs.
Some of the symptoms which can be shown by the child as they grow with Stoneman Syndrome:

- In initial stages movement of joints are restricted, especially neck and shoulders.
- Spinal deformity.
- Frequently imbalance occurs while moving is followed by low-grade fever, inflammation, and joint pain.
- Deformation occurs in bone growth throughout the body (except the tongue, diaphragm, extraocular muscles, cardiac muscle, and smooth muscle).
- Difficulty occurs in breathing, eating, and speaking.
- Hearing loss (rare).
- Swelling occurs in the affected area.

The above symptoms will get more severe as a child grows, tissues not only turn into bones in the upper region of the body, they make their way downwards to the trunk, back, hips, and limbs until the person gets completely immobilized. [6]

DIAGNOSIS

It is a challenging task to diagnose rare genetic diseases. At initial stages, cases of FOP i.e. Fibrodysplasia ossificans progressiva or Stoneman syndrome can be confused with other aggressive muscular illnesses like juvenile fibromatosis, cancer, and fibrous dysplasia. Abnormalities of Big toes is the only symptoms which make it different from other conditions.

In medical diagnosis, the healthcare professional will examine your medical and your family medical history for identifying any genetic pattern, followed by a series of physical examinations and laboratory tests. The laboratory test may include imaging tests such as X-ray, MRI and a genetic testing registry (GTR).

Imaging tests help the doctor to see the overall growth of the heterotopic bone whereas GTR will help the doctor to choose appropriate genetic tests for this condition. GTR is conducted by a group of medical professionals that majorly deals with research and analysis of such rare conditions and it includes a questionnaire for both patient and the medical team. [6]

TREATMENT

Fibrodysplasia ossificans progressiva do not have an effective cure till now, and since the removal of the bone will only lead to the growth of new and more painful bone formation, medical professionals have come up with a couple of medications that slow down the growth and intensity of ossification, they are as follow: -

- Corticosteroids like Prednisone to reduce pain and sudden swelling seen in the early stages. It is recommended to be within 24hrs of the first outburst, before. NSAIDS are to be given between sudden swelling
- Drugs that relax muscles.
- Mast cell inhibitors.
- Aminobisphosphonates.
- Devices such as braces or special shoes to help with walking.
- Occupational therapy.

Those things are to be monitored that can increase the ossification and physical discomfort.

Treatments and medical diagnoses associated with bone such as biopsy, Intramuscular injections (immunizations) or injections, and jaw stretching during dental procedures must be avoided, as it can damage the skeletal system which can speed up the process of ossification in the affected area.

FOP is dangerous for pregnant women, childbirth must be avoided due to two reasons, firstly the genetic malfunction can be transferred to the baby which will make the child's survival difficult. Secondly, the increasing fetus in your womb can increase the risk of sudden outbursts which can be life-threatening for both baby and the mother. [6]

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