Mini-Review

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Stoneman Syndrome: Where Flesh Turns to Bone

Rakshith Balaji[1], **Akshaya N Shetti**[2*]

[1]Grade 12, The Indian High School, Dubai, UAE

[2]Department of Anaesthesiology and critical care, DBVPRMC, PIMS, Loni, Maharashtra, India

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***Corresponding author:** Shetti AN, Department of Anaesthesiology and critical care, DBVPRMC, PIMS, Loni, Maharashtra, India, Orcid Id- 0000-0002-4688-8071, Email: aksnsdr@gmail.com

A B S T R A C T

Fibrodysplasia Ossificans Progressiva (FOP), also known as StoneMan syndrome, is a rare autosomal dominant genetic disorder characterized by abnormal bone development in non-skeletal areas, causing significant suffering. Due to its rarity, awareness and understanding are lacking, leading to delayed diagnoses. This article aims to raise awareness for early detection and treatment, sparking further research.

Keywords: Fibrodysplasia; Ossificans; Progressiva; Stone man; Genetic

Short Review

Fibrodysplasia Ossificans Progressiva (FOP) or StoneMan syndrome is a rare autosomal dominant genetic disorder characterised by the abnormal development of bone in areas of the body where bone is not normally present, hence causing a lot of misery for those affected. Due to its rarity, there is a lack of awareness and knowledge on this disease, leading to a faulty or late diagnosis. Very little is understood about the disease, which can make it challenging to effectively administer analgesia to FOP patients. This article is aimed at spreading awareness of this disease, to ensure an early diagnosis and provide necessary treatment to minimise suffering for those affected and possibly ignite further research into its mechanism and treatment.

Aside from being inherited, FOP can be caused by a mutation in gene 2q23-24 which encodes a bone morphogenetic protein receptor called Activin Receptor Type IA (ACVR1)[1]. The presence of bone morphogenetic protein (BMP) receptors on the surface of cells plays a role in deciding the fate of the stem cells. More specifically, FOP results in the specific substitution of arginine, at position 206 in the ACVR1 protein for histidine. A mutation in such an important receptor changes the fate of stem cells, for example, the cells which were supposed to develop into and grow as soft tissue, may end up ossifying and becoming osseous tissue, which is the characteristic trait of FOP.

The first signs of FOP are congenital deformations in the toes. Heterotopic ossification initially begins with firm and tender swellings on specific body parts such as the back, neck, and shoulders. These swellings cause pain and stiffness and eventually turn into mature bone as they shrink. While this abnormal bone growth can happen without any specific cause, it often occurs after a soft tissue injury or a viral illness[2]. Furthermore, Swelling may occur due to obstruction of tissue fluid by abnormal bone formation. Affected joints may have severely restricted motion causing immobilisation, causing them to require a wheelchair by their late 20s. The individual may also have kyphosis or scoliosis and may exhibit an increased susceptibility to respiratory infection or right sided congestive heart failure. Some cases have even shown hearing impairment, hair loss and mild cognitive delay[3]. Those with FOP have three times higher probability of developing renal stones than the general population. Nausea, swallowing difficulty, vomiting, gastroesophageal reflux, etc further reduce their quality of life[4].

There isn't yet a permanent cure but courses of high-dose corticosteroids at the start of a flare-up can reduce some of the symptoms of the condition. Affected persons may participate in occupational therapy and palliative care can be administered. They must avoid any physical trauma and must take measures to avoid viral illnesses to prevent the symptoms from spreading or worsening. Furthermore, subcutaneous injections should be preferred over intramuscular injections.

This disease affects roughly one in two million people and can become severely disabling. People with FOP have a median lifespan of 40 years. There are around 900 known patients out of an estimated 4000 affected individuals globally. This disorder affects both males and females from all ethnic backgrounds[5].

Due to its low prevalence, there is a dearth of knowledge about the disease as well as the paucity of information available about its treatment. FOP is an illness that causes a great deal of distress for people, the different symptoms linked with the ailment decrease quality of life, making it even worse and necessitating substantial studies to learn more about this health issue. Hence, there is a dire need to work on the development of effective treatments for fibrodysplasia ossificans progressiva.

Conflicts of Interest

This study does not have any conflict of interest.

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