

A Small Clinical Approach to Skeletal Dysplasia

Rashmi Sood*

Department of Immunohematology and Transfusion Medicine, SCH, New Delhi, India

Abstract

Skeletal dysplasia is a type of genetic disorder in which skeleton of the body i.e. bones, joints is highly affected causing problem in children's growth and development. In this case, generally it cause abnormally shaped bones of the legs and arms. Patients with skeletal dysplasia are often have relatively small limbs compared to the other part of the body. It is a kind of genetic disease. It is also known as osteochondrodysplasias.

Keywords: Skeletal dysplasia; Abnormal; Genetic; Thrombosis; Health; Treatment

Introduction

Skeletal dysplasia is a group of disorder due to genetic reason; sometimes transformation of the mutant gene from parents to children occurs because of which paediatric skeletal dysplasia occurs. Because of which abnormalities of cartilage and bone growth happens. The molecular basis for a large majority of these disorders is now known. Commonly seen skeletal dysplasia include achondroplasia, osteogenesis imperfecta, thanatophoric dysplasia, campomelic dysplasia, and hypochondroplasia. It also can be accompanied by involvement of the neurologic, respiratory, and cardiac systems like other systems. Generally, they differ in natural genetic histories and prognoses.

Skeletal dysplasia can lead to Difficulty breathing, some cases breathing stops for 20 seconds or more, Spinal problems including curving, bowing, or narrowing of the spine, Fluid build-up around the brain, Obesity as well as Chronic ear infections etc. Defective genes that either are inherited from a parent or mutate randomly during fetal development cause this disorder. There are so many types of dysplasia such as Achondroplasia (most common) and dwarfism, Osteogenesis imperfecta, Thanatophoric dysplasia which cause extremely short limbs. Hypochondroplasia resulting short arms and legs, and short, broad hands and feet, Campomelic dysplasia resulting bowed long bones in legs and arms, often fatal for new borns and Achondrogenesis resulting skeletal abnormalities including short limbs and a small body.

Skeletal dysplasia is often noticeable at birth, but sometimes it may not develop until later in childhood. It includes slow growth, large head, especially the forehead, short upper arms and thighs, too many fingers or toes or short fingers and toes, arthritis joint pain, Curved bone especially spine, crowded teeth, late walking type of developmental delays, Mental retardation etc.

Diagnosis can easily occur by detailed observation but sometimes for confirmation Doctor suggests X-ray, MRI, CT scans, precisely examine bones, Parental diagnosis imaging such as ultrasound as well as parental genetic testing in order to identify the mutation of gene with a family history of skeletal dysplasia.

Depending upon the condition of the patient the treatment happens such as growth hormone treatment for patient suffering from dwarfism related to deficiency of growth hormone, to improve muscle strength physical therapy, back braces for improvement of spine bowing, orthodontic treatment for teeth, to treat obesity guidance of proper nutrition and exercise.

Conclusion

In some extreme cases, surgical treatment also helps such as in order to treat the chronic ear infection insertion of draining tube in the middle ear, improving breathing by removing tonsils, to relieve the pressure on spinal cord the widening of spinal cord, by draining the excess fluid from brain.

Correspondence to: Rashmi Sood, Department of Immunohematology and Transfusion Medicine, SCH, New Delhi, India, **Tel: 8908441401**, Email: rsood@maw.com

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