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Genetic Underpinnings of Adolescent Idiopathic Scoliosis: A Review

Shipra[1], Hemender Singh[1], Vinod Singh[1], Ekta Rai[1, 2, *] and Swarkar Sharma[3, *]

[1]Human Genetics Research Group, School of Biotechnology, Shri Mata Vaishno Devi University, Katra, Jammu and Kashmir, India.
[2]School of Life Sciences, Jawaharlal Nehru University, New Delhi, India
[3]Centre for Molecular Biology, Central University of Jammu, Jammu and Kashmir, India
E-mail: <shipra.kowra@gmail.com>, <hemender.singh4737@gmail.com>
<vinod.singh@smvdu.ac.in>, ekta.rai@smvdu.ac.in>, <swarkar.molb@cujammu.ac.in>

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ABSTRACT Adolescent Idiopathic Scoliosis (AIS) is a three-dimensional spine deformity with lateral curvature having a Cobb angle exceeding 10° in the individual. It affects about 1-4 percent of adolescents globally and more frequently occurs in females than males. Despite the extensive research carried out on AIS, its aetiology is not known yet. However, several genetic studies suggest the contribution of various genetic variants in the possible aetiology of AIS. This review summarises the genetic association studies, including linkage, candidate as well as genome-wide association studies that were carried out globally on AIS and also categorised the associated genes in different biological pathways such as neurodevelopmental, hormone-related, cartilage and bone development pathways, based on their potential functional roles in the respective pathway, to understand the pathology of the disorder.