

Variations in exon-2 of SBDS gene and its association with aplastic anemia

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Aplastic anemia is more prevalent in Asia than the West, with majority of cases being of acquired rather than constitutional etiology. Telomere shortening is a feature common to both Schwachman-Diamond syndrome and aplastic anemia, making the *SBDS* gene associated with Schwachman-Diamond syndrome a candidate for aplastic anemia. Telomeres shortening in only one third of the aplastic anemia patients were found to be linked with *TERC* and *TERT* gene mutations. The facts that, the number of aplastic anemia patients exceeds the number of cases with telomerase complex mutations and presence of *SBDS* mutation in acquired aplastic anemia make this gene a candidate for association with aplastic anemia. The presence of this mutation has not been investigated in India. We performed a case-control study that is aimed at comparing the proportions of patients and control presenting with two most common gene conversion mutations of the *SBDS* gene, viz. 258+2T>C and 183-184TA>CT in north Indian aplastic anemia patients. In the present study, no mutations of the *SBDS* gene were detected; however, two silent nucleotide changes were seen with a higher frequency than in Japanese and European populations. We conclude that gene conversions of *SBDS* gene do not contribute to precipitation of aplastic anemia.

Biography

Dharmendra Jain has completed his post-graduation in Biotechnology in the year, 2004 and submitted his thesis for the award of Doctorate of Philosophy (Ph.D.). He has a total of ten years of experience in the field of molecular genetics and immunology, in clinical settings. He has been able to publish a total of nine national and international publications in leading journals of repute. The area of interest is molecular cytogenetic, transplant immunology, immunophenotyping and auto immune diagnosis. The research work is oriented towards the diagnosis and management of aplastic anemia.

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