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Geographic Isolation and Endogamous Practices Provide Higher Risk of Genetic Disorders in Jammu and Kashmir

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Abstract

Rare disorders are poorly understood, most often remain uncharacterized or patients are misdiagnosed due to lack of specific clinical resources. Understanding the basics of inheritance is essential in such cases as it helps to figure out the plausibility of a disorder as an inherited or genetic disease. Though identification and characterization of such disorders is complicated, Next generation Sequencing has come up as a tool in recent times and is of great help. It is quite visible in literature that since the advent of this methodology, a drastic increase in identification and genetic characterization of various rare diseases across the world has occurred. We emphasize on NGS/WES, as an effective method in understanding uncharacterized Mendelian Disorders. It is of great help, especially in developing countries and regions like Jammu and Kashmir where, such familial disorders exist in abundance, due to very high consanguinity, but remain undiagnosed/misdiagnosed due to lack of specialized testing. We have collected huge number of highly extended families representing various rare genetic disorders and trying to elucidate the genetic cause and biology of the diseases in these families.

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