



Category: Miscellaneous

Promises and pitfalls of NGS technology in Clinic: Experience from Kolkata

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Abstract

The NGS technology has revolutionized clinical diagnostics in India starting from 2013 onwards. With the continuous decline of sequencing cost NGS based tests is becoming affordable to a large section of the masses. However medical education has not been able to keep pace with genomic technological advancements leading to a gap in the knowledgebase of practicing clinicians. The current work stresses the clinical challenges encountered while delivering genomic solutions to clinical problems in Eastern India specifically in the city of Kolkata.

With the help of a series of cases the utility of NGS testing and different flavors of the technology will be outlined. Specifically in the prenatal diagnostic setting dilemma related to testing and not testing will be outlined. The absence of customized CNV assays complimenting the NGS panel will be highlighted.

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